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MOST RECENT UPDATE:           200879      <200879/DW>
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    and 20061231/UPIC, 20070601/UPIC, 20071001/UPIC, 20071130/UPIC,
    20080401/UPIC, 20080701/UPIC and 20081001/UPIC.
    ECLA reclassifications to mid August and US national classification
    mid September 2008 have also been loaded. Update dates 20080401,
    20080701 and 20081001/UPEC and /UPNC have been assigned to these. <<

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>>> HELP for European Patent Classifications see HELP ECLA, HELP ICO <<<



=> d l67 que
L8              QUE  ABB=ON  PLU=ON  DETERMIN? OR IDENTIF? OR DIAGNOS? OR
                 DETECT?
L9              QUE  ABB=ON  PLU=ON  SCREEN?
L11             QUE  ABB=ON  PLU=ON  FETUS
L13             QUE  ABB=ON  PLU=ON  CHROMOSOM?(2A)ABNORMAL?
L14             QUE  ABB=ON  PLU=ON  DOWN(2A)SYNDROME?
L17             QUE  ABB=ON  PLU=ON  MARKER? OR INDICAT!R?
L18             QUE  ABB=ON  PLU=ON  PARAMETER? OR VALUE
L22             QUE  ABB=ON  PLU=ON  (PREGNAN? OR FETUS)(3A)(L13 OR L14)
L32         307 SEA FILE=WPIX ABB=ON  PLU=ON  (L8 OR L9)(3A)(L13 OR L14)

L33          49 SEA FILE=WPIX ABB=ON  PLU=ON  L32 AND L11
L34          23 SEA FILE=WPIX ABB=ON  PLU=ON  L33 AND (L17 OR L18)
L35          18 SEA FILE=WPIX ABB=ON  PLU=ON  L34 AND L22
L36          18 SEA FILE=WPIX ABB=ON  PLU=ON  L35 AND (PY<=2006 OR
                PRY<=2006 OR AY<=2006)
L42             QUE  ABB=ON  PLU=ON  PROBABILIT?
L43             QUE  ABB=ON  PLU=ON  STATISTIC?
L67           4 SEA FILE=WPIX ABB=ON  PLU=ON  L36 AND (L42 OR L43)


=> d l67 ifull 1-4

L67  ANSWER 1 OF 4  WPIX COPYRIGHT 2008       THOMSON REUTERS on STN
ACCESSION NUMBER:     2005-385944 [39]   WPIX
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CROSS REFERENCE: 2007-475698
DOC. NO. CPI: C2005-119285 [39]
DOC. NO. NON-CPI: N2005-313084 [39]
TITLE: Prenatal diagnosis and/or screening of genetic disorders and congenital abnormalities, including Down syndrome, Hemophilia, Wilms tumor and muscular atrophy, using array-based hybridization of cell-free fetal DNA from amniotic fluid
DERWENT CLASS: B04; D16; P31; S03; S05; T01
INVENTOR: BIANCHI D; BIANCHI D W; LARRABEE P; LARRABEE P B; LESHANE E; LESHANE E S; JOHNSON K L
PATENT ASSIGNEE: (TUFT-N) TUFTS-NEW ENGLAND MEDICAL CENT; (BIAN-I) BIANCHI D W; (JOHN-I) JOHNSON K L; (LARR-I) LARRABEE P B
COUNTRY COUNT: 107

PATENT INFORMATION:

PATENT NO	KIND	DATE	WEEK	LA	PG	MAIN IPC
WO 2005044086 <--	A2	20050519	(200539)*	EN	110[7]	
EP 1678329 <--	A2	20060712	(200648)	EN		
AU 2004286845 <--	A1	20050519	(200681)	EN		
JP 2007515947	W	20070621	(200742)	JA	74	
US 20070212689	A1	20070913	(200761)	EN		

APPLICATION DETAILS:

PATENT NO	KIND	APPLICATION	DATE
WO 2005044086 20041029	A2	WO 2004-US35929	
AU 2004286845 20041029	A1	AU 2004-286845	
EP 1678329 20041029	A2	EP 2004-818325	
EP 1678329 20041029	A2	WO 2004-US35929	
JP 2007515947 20041029	W	WO 2004-US35929	
JP 2007515947 20041029	W	JP 2006-538287	
US 20070212689 20031030	A1 Provisional	US 2003-515735P	
US 20070212689 20041029	A1	WO 2004-US35929	
US 20070212689	A1	US 2007-577341	20070214

FILING DETAILS:

PATENT NO	KIND		PATENT NO	
EP 1678329	A2	Based on	WO 2005044086	A
AU 2004286845	A1	Based on	WO 2005044086	A
JP 2007515947	W	Based on	WO 2005044086	A

PRIORITY APPLN. INFO: US 2003-515735P 20031030

US 2007-577341 20070214
INT. PATENT CLASSIF.:
IPC ORIGINAL: C12N0015-09 [I,A]; C12N0015-09 [I,C]; C12Q0001-68 [I,A]; C12Q0001-68 [I,C]; G01N0021-77 [I,C]; G01N0021-78 [I,A]; G01N0033-50 [I,A]; G01N0033-53 [I,A]; G01N0033-53 [I,C]; G01N0033-58 [I,A]; G01N0033-58 [I,C]; G01N0037-00 [I,A]; G01N0037-00 [I,C]
IPC RECLASSIF.: A61B [I,S]; C12Q0001-68 [I,A]; C12Q0001-68 [I,C]
USCLASS NCLM: 435/006.000
BASIC ABSTRACT:

WO 2005044086 A2 UPAB: 20051222
NOVELTY - Prenatal diagnosis comprising providing a sample of amniotic fluid fetal DNA, analyzing the amniotic fluid fetal DNA by hybridization to obtain fetal genomic information and based on the fetal genomic information obtained, providing a prenatal diagnosis, is new.
DETAILED DESCRIPTION - INDEPENDENT CLAIMS are also included for:
(1) testing amniotic fluid fetal DNA by array-based comparative genomic hybridization, comprising providing a test sample of amniotic fluid fetal DNA, where the test sample comprises a plurality of nucleic acid segments comprising a substantially complete first genome with a chromosomal microabnormality and labeled with a first detectable agent, providing a reference sample of control genomic DNA, where the reference sample comprises a plurality of nucleic acid segments comprising a substantially complete second genome with a known karyotype and labeled with a second detectable agent, providing an array comprising a plurality of genetic probes, where each genetic probe is immobilized to a discrete spot on a substrate surface to form the array and where together the genetic probes comprise a substantially complete third genome or a subset of a third genome, contacting the array simultaneously with the test sample and reference sample under conditions where the nucleic acid segments of the test and reference samples can specifically hybridize to the genetic probes immobilized on the array, using a computer-assisted imaging system capable of acquiring multicolor fluorescence images to obtain a fluorescence image of the array after hybridization, using a computer-assisted image analysis system to analyze the fluorescence image obtained, to interpret data imaged from the array and to display results as genome copy number ratios as a function of genomic locus in the third genome, determining the karyotype of the first genome by FISH analysis, and comparing the results displayed as genome copy number ratios to the karyotype of the first genome determined by FISH;
(2) identifying a chromosomal abnormality by analyzing amniotic fluid fetal DNA by array-based comparative genomic hybridization, comprising providing a test sample of amniotic fluid fetal DNA, where the amniotic fluid fetal DNA originates from a fetus determined to have multiple congenital anomalies by sonographic examination, where the test sample comprises a plurality of nucleic segments comprising a substantially complete and acid first genome with a normal karyotype and labeled with a first detectable agent, providing a reference sample of control amniotic fluid fetal DNA, where the control amniotic fluid fetal DNA originates from a fetus determined to have no congenital anomalies by sonographic examination, and where the reference sample comprises a plurality of nucleic acid segments comprising a substantially complete second genome with a normal karyotype and labeled with a second detectable agent, providing an array comprising a plurality of genetic probes, where each genetic probe is immobilized to a discrete spot on a substrate surface to form the array and wherein together the genetic probes comprise a substantially complete third genome or a subset of a third genome, contacting the array simultaneously with the test sample and reference sample under conditions where the nucleic acid segments in the samples can specifically hybridize to the genetic probes immobilized on the array, using a computer-assisted imaging system capable of acquiring multicolor fluorescence

images to obtain a fluorescence image of the array after hybridization, using a computer-assisted image analysis system to analyze the fluorescence image obtained, to interpret data imaged from the array and to display results as genome copy number ratios as a function of genomic locus in the third genome, and analyzing the results displayed to detect and identify any chromosomal abnormality present; and

(3) a kit comprising materials to extract cell-free fetal DNA from a sample of amniotic fluid obtained from a pregnant woman, an array comprising a plurality of genetic probes, where each genetic probe is immobilized to a discrete spot on a substrate surface to form the array and where together the genetic probes comprise a substantially complete genome or a subset of a genome, and instructions for using the array in the methods mentioned above.

USE - The methods and compositions of the present invention are useful for the prenatal diagnosis, screening, monitoring and/or testing of genetic disorders and congenital abnormalities, including Down syndrome, Patau syndrome, Edward syndrome, Turner syndrome, Klinefelter syndrome or XYY disease, Hemophilia A, Duchenne muscular dystrophy, LeschNyhan syndrome, severe combined immunodeficiency, Fragile X syndrome, Prader-Willi syndrome, Angelman syndrome, DiGeorge syndrome, Smith-Magenis syndrome, Rubinstein-Taybi syndrome, Miller-Dieker syndrome, Williams syndrome, Charcot-Marie-Tooth syndrome, Cri du Chat syndrome, Retinoblastoma, Wolf-Hirschhorn syndrome, Wilms tumor, muscular atrophy, cystic fibrosis, Gaucher disease, Marfan syndrome, sickle cell anemia and spinobulbar muscular atrophy (all claimed). TECHNOLOGY FOCUS:

BIOTECHNOLOGY - Preferred Method: The amniotic fluid fetal DNA in the prenatal diagnosis is obtained by providing a sample of amniotic fluid obtained from a woman pregnant with a fetus , removing cell populations from the sample of amniotic fluid to obtain a remaining amniotic material, and treating the remaining amniotic material such that cell-free fetal DNA present in the remaining material is extracted and made available for analysis, resulting in amniotic fluid fetal DNA. Substantially all cell populations are removed from the sample of amniotic fluid and where the amniotic fluid fetal DNA consists essentially of cell-free fetal DNA. The remaining amniotic material comprises some cells, where the amniotic fluid fetal DNA comprises cell-free fetal DNA and DNA originating from the cells present in the remaining amniotic material. The method further comprises freezing the remaining amniotic material to obtain a frozen sample, storing the frozen sample for a period of time under suitable storage conditions, and thawing the frozen sample prior to the treating step, and removing substantially all cell populations that are still present in the remaining amniotic material after the thawing step and prior to the treating step. Analyzing the amniotic fluid fetal DNA by hybridization to obtain fetal genomic information comprises using an array that is a cDNA array, an oligonucleotide array or a SNP array, or is performed using array-based comparative genomic hybridization. The method also comprises amplifying the amniotic fluid fetal DNA prior to the analyzing step, resulting in amplified amniotic fluid fetal DNA, where amplifying the amniotic fluid fetal DNA comprises using PCR. The method also comprises labeling the amniotic fluid fetal DNA with a detectable agent prior to the analyzing step, resulting in labeled amniotic fluid fetal DNA, where the detectable agent comprises a fluorescent label that comprises a fluorescent dye selected from Cy-3, Cy-5, Texas Red, FITC, Spectrum Red, Spectrum Green, phycoerythrin, a rhodamine, a fluorescein, a fluorescein isothiocyanate, a carbocyanine, a merocyanine, a styryl dye, an oxonol dye, a BODIPY dye, their equivalents, analogues, derivatives, or their combination. Labeling the amniotic fluid fetal DNA comprises random priming, nick translation, PCR or tailing. The detectable agent comprises biotin

or dioxigenin. The fetal genomic information includes chromosomal abnormalities and genome copy number changes at multiple genomic loci.

Providing a prenatal diagnosis comprises determining the sex of the fetus, detecting and identifying a chromosomal abnormality, and identifying a disease or condition associated with a chromosomal abnormality. The fetus is suspected of having a chromosomal abnormality, and of having a disease or condition associated with a chromosomal abnormality. The pregnant woman is 35 or more than 35 years old. The chromosomal abnormality is an extra individual chromosome, a missing individual chromosome, an extra portion of a chromosome, a missing portion of a chromosome, a break, a ring, a chromosomal rearrangement, or their combination, a chromosomal rearrangement selected from the group consisting of a translocation, an inversion, a duplication, a deletion, an addition, or their combination, an extra chromosome 21, a missing chromosome 21, an extra portion of chromosome 21, a missing portion of chromosome 21, a rearrangement of chromosome 21, or their combination, not detectable by G-banding analysis or metaphase CGH, is a microdeletion, a microduplication, or a subtelomeric rearrangement, and/or is an extra chromosome 13, 18, X or Y, a chromosomal aberration involving chromosome 1, a deletion of chromosome portion 1q21, a deletion of chromosome portion 4p16, a chromosomal aberration involving chromosome 4, a deletion on chromosome 5, a chromosomal aberration involving chromosome 7, a deletion of chromosome portion 7q11.23, a chromosomal aberration involving chromosome 8, a translocation involving chromosome 9 and chromosome 22, a chromosomal aberration involving chromosome 10, a chromosomal aberration involving chromosome 11, a deletion of chromosome portion 13q14, a deletion of chromosome portion 15q11-q13, a deletion of chromosome portion 15q21.1, a deletion of chromosome portion 16p13.3, a deletion of chromosome portion 17pl 1.2, a deletion of chromosome portion 17p13.3, a chromosomal aberration involving chromosome19, a deletion of chromosome portion 22q11, and a chromosomal aberration involving chromosome X. The disease or condition associated with a chromosomal abnormality is an aneuploidy that is Down syndrome, Patau syndrome, Edward syndrome, Turner syndrome, Klinefelter syndrome or XYY disease, and/or associated with a chromosomal abnormality is an X-linked disorder that is Hemophilia A, Duchenne muscular dystrophy, LeschNyhan syndrome, severe combined immunodeficiency, or Fragile X syndrome, and/or associated with a chromosomal abnormality that is not detectable by G-banding analysis or metaphase CGH, and/or associated with a chromosomal abnormality is a microdeletion/microduplication syndrome, such as Prader-Willi syndrome, Angelman syndrome, DiGeorge syndrome, Smith-Magenis syndrome, Rubinstein-Taybi syndrome, Miller-Dieker syndrome, Williams syndrome, and Charcot-Marie-Tooth syndrome. The disease or condition is also associated with a subtelomeric rearrangement, such as Cri du Chat syndrome, Retinoblastoma, Wolf-Hirschhorn syndrome, Wilms tumor, muscular atrophy, cystic fibrosis, Gaucher disease, Marfan syndrome, sickle cell anemia and spinobulbar muscular atrophy.

The method alternatively comprises analyzing amniotic fluid fetal DNA by array-based comparative genomic hybridization, comprising providing a test sample of amniotic fluid fetal DNA, where the test sample comprises a plurality of nucleic acid segments comprising a substantially complete first genome with an

unknown karyotype and labeled with a first detectable agent, providing a reference sample, where the reference sample comprises a plurality of nucleic acid segments comprising a substantially complete second genome with a known karyotype and labeled with a second detectable agent, providing an array comprising a plurality of genetic probes, where each genetic probe is immobilized to a discrete spot on a substrate surface to form the array and where together the genetic probes comprise a substantially complete third genome or a subset of a third genome, contacting the array simultaneously with the test and reference samples under conditions wherein the nucleic acid segments in the samples can specifically hybridize to the genetic probes on the array, determining the binding of the individual nucleic acids of the test sample and reference sample to the individual genetic probes immobilized on the array to obtain a relative binding pattern, and based on the relative binding pattern obtained, providing a prenatal diagnosis.

Determining the binding of the individual nucleic acids of the test and reference probes immobilized on the array to comprise samples to the individual genetic obtain a relative binding pattern measuring the intensity of the signals produced by the first detectable agent and second detectable agent at each discrete spot on the array; and determining the ratio of the intensities of the signals for each spot of the array.

Determining the binding of the individual nucleic acids of the test and reference samples to the individual genetic probes immobilized on the array to obtain a relative binding pattern comprises using a computer-assisted imaging system capable of acquiring multicolor fluorescence images to obtain a fluorescence image of the array after hybridization, and using a computer-assisted image analysis system to analyze the fluorescence image obtained, to interpret data imaged from the array and to display results as genome copy number ratios as a function of genomic locus in the third genome.

Providing a prenatal diagnosis comprises determining the sex of the fetus carried by the pregnant woman, detecting and identifying a chromosomal abnormality, and identifying a disease or condition associated with a chromosomal abnormality. The amniotic fluid fetal DNA originates from a fetus suspected of having a chromosomal abnormality, from a fetus suspected of having a disease or condition associated with a chromosomal abnormality, or has been extracted from a sample of amniotic fluid obtained from a pregnant woman who is 35 or more than 35 years old. The nucleic acids of the test sample and reference sample in any of the methods cited are labeled by random priming, nick translation, PCR or tailing. The first detectable agent comprises a first fluorescent label and the second detectable agent comprises a second fluorescent label. The first fluorescent label and second fluorescent label produce a dual-color fluorescence upon excitation. The first fluorescent label also comprises Cy-3 or Spectrum Red and the second fluorescent label comprises Cy-5 or Spectrum Green, and/or the first fluorescent label comprises Cy-5 or Spectrum Green and the second fluorescent label comprises Cy-3 or Spectrum Red. The hybridization capacity of high copy number repeat sequences present in the nucleic acid segments of the test sample and reference sample is suppressed by adding unlabeled blocking nucleic acids to the test sample and reference sample prior to the contacting step. The unlabeled blocking nucleic acids are Human Cot-1 DNA. The amniotic fluid fetal DNA is obtained by providing a sample of amniotic fluid

obtained from a woman pregnant with a fetus, removing cell populations from the sample of amniotic fluid to obtain a remaining amniotic material, and treating the remaining amniotic material such that cell-free fetal DNA present in the remaining material is extracted and made available for analysis, resulting in amniotic fluid fetal DNA. Substantially all cell populations are removed from the sample of amniotic fluid, where the amniotic fluid fetal DNA consists essentially of cell-free fetal DNA. The remaining amniotic material comprises some cells and where the amniotic fluid fetal DNA comprises cell-free fetal DNA and DNA originating from the cells present in the remaining amniotic material.

The method further comprises freezing the remaining amniotic material to obtain a frozen sample, storing the frozen sample for a period of time under suitable storage conditions, and thawing the frozen sample prior to the treating step, amplifying the amniotic fluid fetal DNA using PCR, resulting in amplified amniotic fluid fetal DNA, and labeling the amniotic fluid fetal DNA with a detectable agent by random priming, nick translation, PCR or tailing, resulting in labeled amniotic fluid fetal DNA. The karyotype of the second genome has been determined by G-banding analysis, metaphase CGH, FISH or SKY. Comparing the results displayed as genome copy number ratios to the karyotype of the first genome determined by FISH in testing amniotic fluid fetal DNA by array-based comparative genomic hybridization comprises evaluating the degree of consistency between the results displayed and the karyotype of the first genome determined by FISH and/or by array-based hybridization. The chromosomal micro-abnormality is a microdeletion, a microduplication or a subtelomeric rearrangement, where the micro-abnormality is a deletion of chromosome portion 1q22, a deletion of chromosome portion 7q11.23, a deletion of chromosome portion 8q21, a deletion of chromosome portion 10q21.1-q22.1, a deletion of chromosome portion 15q11-q13, a deletion of chromosome portion 16p13.3, a deletion of chromosome portion 17p 11.2, a deletion of chromosome portion 17p13.3, a deletion of chromosome portion 19q13.1-q13.2, or a deletion of chromosome portion 22q11.2. The karyotype of the test sample in identifying a chromosomal abnormality by analyzing amniotic fluid fetal DNA by array-based comparative genomic hybridization has been determined by metaphase CGH analysis with a 550 band level of resolution. The chromosomal abnormality present in the first genome is a chromosomal micro-abnormality that is not detectable by metaphase CGH analysis with a 550 band level of resolution, and is selected from a micro-addition, a micro-deletion, a micro-duplication, a micro-inversion, a micro-translocation, a subtelomeric rearrangement and their combination. The test and reference samples are matched for fetal gender, site of sample acquisition, gestational age and storage time.

Preferred Kit: The kit further comprises materials to label a first sample of DNA with a first detectable agent and a second sample of DNA with a second detectable agent. The first detectable agent comprises a first fluorescent label, the second detectable agent comprises a second fluorescent label, and the first and second fluorescent labels produce a dual-color fluorescence upon excitation. The kit also comprises materials to label a first sample of DNA and a second sample of DNA with Cy-3 and Cy-5, and/or Spectrum Red and Spectrum Green. The kit also comprises a sample of control genomic DNA with a normal, female or male karyotype, or with a karyotype comprising a chromosomal abnormality, and

hybridization and wash buffers, and Human Cot-1 DNA.

EXTENSION ABSTRACT:

EXAMPLE - Frozen amniotic fluid supernatant specimens were obtained from the Tufts-New England Medical Center (Tufts-NEMC) Cytogenetics Laboratory. All samples were collected for routine indications, such as advanced maternal age, abnormal maternal serum screening results, or detection of a fetal sonographic abnormality. Real-time quantitative PCR analysis was performed using a Perkin-Elmer Applied Biosystems (PE-ABI) 7700 Sequence Detector. Analysis was based on the 5'-to-3' exonuclease activity of the Tap DNA polymerase, using the FCY locus as a basis for detecting male DNA if the fetus was male. The FCY primers were derived from the Y-chromosome-specific sequence Y49a. In 21 samples, the known fetal karyotype was 46, XX (normal female), in 15 samples the known fetal karyotype was 46, XY (normal male), and in two samples, the known karyotype was 47, XY, +21 (male fetus with Down syndrome). The samples were coded and analyzed blindly. In the female fetuses 0 GE/mL of DYSI DNA was detected in the amniotic fluid. The mean value of DYSI DNA detected in male fetuses was 2,668 GE/mL. Linear regression analysis showed a correlation between fetal DNA and gestational age. In all 38 cases, the predicted fetal gender was correct. The results were statistically significant. In the cases of fetal Down syndrome, there was no elevation of the amount of fetal DNA compared to the samples obtained from fetuses with a normal male karyotype.

FILE SEGMENT: CPI; GMPI; EPI

MANUAL CODE: CPI: B01-D02; B04-B03C; B04-B04L; B04-E03; B04-E05; B06-H; B11-C07B3; B11-C08E3; B11-C08E5; B11-C08E6; B11-C11; B12-K04A3; B12-K04F; D05-H09; D05-H10; D05-H18B
EPI: S03-E04D; S03-E14H; S05-C; T01-J06A; T01-J13A

L67 ANSWER 2 OF 4 WPIX COPYRIGHT 2008 THOMSON REUTERS on STN

ACCESSION NUMBER: 2005-173182 [18] WPIX

DOC. NO. CPI: C2005-055747 [18]

DOC. NO. NON-CPI: N2005-144410 [18]

TITLE: Determining chromosomal abnormality in fetus involves receiving data comprising value of biological parameter from different stages of pregnancy and determining likelihood data

DERWENT CLASS: B04; D16; S05; T01

INVENTOR: WRIGHT D E

PATENT ASSIGNEE: (UYPL-N) UNIV PLYMOUTH

COUNTRY COUNT: 107

PATENT INFORMATION:

PATENT NO	KIND	DATE	WEEK	LA	PG	MAIN IPC
WO 2005015473 <--	A2	20050217	(200518)*	EN	50[7]	
EP 1668553 <--	A2	20060614	(200641)	EN		
US 20070148631	A1	20070628	(200743)	EN		

APPLICATION DETAILS:

PATENT NO	KIND	APPLICATION	DATE
WO 2005015473 A2 20040712		WO 2004-GB3013	
EP 1668553 A2 20040712		EP 2004-743354	

EP 1668553 A2 WO 2004-GB3013
20040712
US 20070148631 A1 WO 2004-GB3013
20040712
US 20070148631 A1 US 2006-565686
20060710

FILING DETAILS:

PATENT NO	KIND		PATENT NO	
EP 1668553	A2	Based on	WO 2005015473	A

PRIORITY APPLN. INFO: GB 2003-17476 20030725
INT. PATENT CLASSIF.:
IPC ORIGINAL: C12Q0001-00 [I,A]; C12Q0001-00 [I,C]
IPC RECLASSIF.: G01N0033-74 [I,A]; G01N0033-74 [I,C]; G01N0033-76 [I,A]; G06F0019-00 [I,A]; G06F0019-00 [I,C]
ECLA: G01N0033-74B; G01N0033-76; G06F0019-00C
ICO: S01N0333:47A6; S01N0333:575
USCLASS NCLM: 435/004.000
BASIC ABSTRACT:
WO 2005015473 A2 UPAB: 20050708
NOVELTY - Determining chromosomal abnormality in fetus involves receiving data comprising value of biological parameter (e.g. marker) from different stages of pregnancy and determining likelihood data.
DETAILED DESCRIPTION - An INDEPENDENT CLAIM is included for a computer system for providing risk data representing likelihood of fetus having chromosomal abnormality .
USE - For determining likelihood of fetus having chromosomal abnormality e.g. Down's syndrome (claimed).
ADVANTAGE - The chromosomal abnormalities are determined with significantly better results than the procedures of Wald, based upon a counter-intuitive recognition. The method has advantages over alternative techniques such as numerical integration in that errors due to the sampling can be quantified statistically and the number of draws can be determined to achieve the desired precision.
TECHNOLOGY FOCUS:
BIOLOGY - Preferred Markers: The biological marker comprises at least one of total human chorionic gonadotropin (hCG), pregnancy associated plasma protein (PAPP), Inhibin-A, alpha-fetoprotein (AFP), unconjugated estriol (uE3) and it is not free beta-LCG.
FILE SEGMENT: CPI; EPI
MANUAL CODE: CPI: B04-E12; B11-C08F1; B11-C11; B12-K04A3; B12-K04F; D05-H09; D05-H12; D05-H18
EPI: S05-D06; T01-J06A; T01-J13A

L67 ANSWER 3 OF 4 WPIX COPYRIGHT 2008 THOMSON REUTERS on STN
ACCESSION NUMBER: 1994-065833 [08] WPIX
CROSS REFERENCE: 1990-254121; 1993-336078; 1994-176282; 1999-404069
DOC. NO. CPI: C1994-029622 [08]
TITLE: Detection of Down's syndrome in foetuses - by detecting high levels of free beta human chorionic gonadotropin in the maternal blood of pregnant women
DERWENT CLASS: B04
INVENTOR: MACRI J N
PATENT ASSIGNEE: (MACR-I) MACRI J N; (JNMA-N) JN MACRI TECHNOLOGIES INC LLC

COUNTRY COUNT: 44

PATENT INFORMATION:

PATENT NO	KIND	DATE	WEEK	LA	PG	MAIN IPC
WO 9403804 <--	A1	19940217	(199408)*	EN	63[17]	
US 5324667 <--	A	19940628	(199425)	EN	29[17]	
AU 9348043 <--	A	19940303	(199426)	EN		
EP 673508 <--	A1	19950927	(199543)	EN		
JP 08503067 <--	W	19960402	(199645)	JA	56[0]	
EP 673508 <--	A4	19970625	(199746)	EN		
AU 689440 <--	B	19980402	(199823)	EN		
JP 2877516 <--	B2	19990331	(199918)	JA	27	
KR 171451 <--	B1	19990501	(200051)	KO		
EP 673508 <--	B1	20021030	(200272)	EN		
DE 69332456 <--	E	20021205	(200304)	DE		
CA 2141668	C	20070102	(200705)	EN		

APPLICATION DETAILS:

PATENT NO	KIND	APPLICATION	DATE
WO 9403804 A1 19930806		WO 1993-US7408	
US 5324667 A CIP of 19890117		US 1989-297481	
US 5324667 A CIP of 19890217		US 1989-311808	
US 5324667 A CIP of 19890508		US 1989-349373	
US 5324667 A CIP of 19890601		US 1989-360603	
US 5324667 A CIP of 19891012		US 1989-420775	
US 5324667 A CIP of 19920414		US 1992-868160	
US 5324667 A 19920807		US 1992-925844	
EP 673508 A4		EP 1993-918683	
AU 9348043 A		AU 1993-48043	19930806
AU 689440 B		AU 1993-48043	19930806
DE 69332456 E 19930806		DE 1993-632456	
EP 673508 A1 19930806		EP 1993-918683	
EP 673508 B1 19930806		EP 1993-918683	
DE 69332456 E		EP 1993-918683	

19930806		
EP 673508 A1		WO 1993-US7408
19930806		
JP 08503067 W		WO 1993-US7408
19930806		
JP 2877516 B2		WO 1993-US7408
19930806		
KR 171451 B1		WO 1993-US7408
19930806		
EP 673508 B1		WO 1993-US7408
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DE 69332456 E		WO 1993-US7408
19930806		
JP 08503067 W		JP 1994-505582
19930806		
JP 2877516 B2		JP 1994-505582
19930806		
KR 171451 B1		KR 1995-700505
19950207		
CA 2141668 C		CA 1993-2141668
19930806		
CA 2141668 C		WO 1993-US7408
19930806		

FILING DETAILS:

PATENT NO	KIND		PATENT NO	
AU 689440	B	Previous Publ	AU 9348043	A
DE 69332456	E	Based on	EP 673508	A
JP 2877516	B2	Previous Publ	JP 8503067	W
AU 9348043	A	Based on	WO 9403804	A
EP 673508	A1	Based on	WO 9403804	A
JP 08503067	W	Based on	WO 9403804	A
AU 689440	B	Based on	WO 9403804	A
JP 2877516	B2	Based on	WO 9403804	A
EP 673508	B1	Based on	WO 9403804	A
DE 69332456	E	Based on	WO 9403804	A
CA 2141668	C	Based on	WO 9403804	A

PRIORITY APPLN. INFO:	US 1992-925844	19920807
	US 1989-297481	19890117
	US 1989-311808	19890217
	US 1989-349373	19890508
	US 1989-360603	19890601
	US 1989-420775	19891012
	US 1992-868160	19920414
INT. PATENT CLASSIF.:		
MAIN:	G01N033-49; G01N033-74	
SECONDARY:	G01N033-493; G01N033-68; G01N033-76	
IPC ORIGINAL:	G01N0033-53 [I,A]; G01N0033-74 [I,C]; G01N0033-76 [I,A]	
IPC RECLASSIF.:	G01N0033-50 [I,A]; G01N0033-50 [I,C]; G01N0033-53 [I,A]; G01N0033-53 [I,C]; G01N0033-68 [I,A]; G01N0033-68 [I,C]; G01N0033-74 [I,A]; G01N0033-74 [I,C]; G01N0033-76 [I,A]	
ECLA:	G01N0033-76	
USCLASS NCLM:	436/518.000	
NCLS:	435/007.900; 435/007.920; 436/065.000; 436/086.000; 436/087.000; 436/510.000; 436/548.000	

BASIC ABSTRACT:
WO 1994003804 A1 UPAB: 20050507 A screening method for determining a pregnant woman's risk of carrying a fetus with Down's syndrome (DS) is claimed comprising measuring the pregnant woman's maternal blood for free beta human chorionic gonadotropin (HCG) during a time period selected from: the first trimester of pregnancy, the second trimester of pregnancy and the third trimester of pregnancy, and comparing the level of free beta HCG to reference values during the time period in (1) pregnant women carrying DS fetuses and (2) pregnant women carrying normal fetuses, where a higher level of free beta HCG is indicative of a higher probability of carrying a fetus with DS. ADVANTAGE - The method scan correctly predict a higher percentage of fetal DS cases, with a lesser false positive rate, than other known methods. Detection efficiency for DS as high as 83% has been achieved.

FILE SEGMENT: CPI
MANUAL CODE: CPI: B04-B04B1; B04-B04D5; B04-J01; B11-C08; B12-K04A

L67 ANSWER 4 OF 4 WPIX COPYRIGHT 2008 THOMSON REUTERS on STN
ACCESSION NUMBER: 1990-254121 [33] WPIX
CROSS REFERENCE: 1994-065833; 1993-336078; 1999-404069; 1994-176282
DOC. NO. CPI: C1990-110079 [21]
DOC. NO. NON-CPI: N1990-196921 [21]
TITLE: Screening for foetus with Down syndrome - by measuring pregnant woman's blood levels of free beta sub-unit of human chorionic gonadotropin
DERWENT CLASS: B04; D16; S03; S05
INVENTOR: MACRI J N
PATENT ASSIGNEE: (MACR-I) MACRI J N; (MACR-I) MACRI TECHNOLOGIES LLC INC J N; (MACR-N) MACRI TECHNOLOGIES LLC J N
COUNTRY COUNT: 28

PATENT INFORMATION:

PATENT NO	KIND	DATE	WEEK	LA	PG	MAIN IPC
WO 9008325 <--	A	19900726	(199033)*	EN	51[14]	
AU 9050936 <--	A	19900813	(199044)	EN		
EP 409956 <--	A	19910130	(199105)	EN		
CN 1047390 <--	A	19901128	(199132)	ZH		
JP 03505128 <--	W	19911107	(199151)	JA		
US 5258907 <--	A	19931102	(199345)	EN	26[14]	
US 5324668 <--	A	19940628	(199425)	EN	25[12]	
EP 666477 <--	A1	19950809	(199536)	EN	32[14]	
EP 409956 <--	B1	19960327	(199617)	EN	32[14]	
DE 69026153 <--	E	19960502	(199623)	DE		
ES 2084689 <--	T3	19960516	(199627)	ES		
JP 2644373 <--	B2	19970825	(199739)	JA	22[0]	

EP 666477 B1 20031015 (200368) EN
<--
DE 69034111 E 20031120 (200401) DE
<--
ES 2210266 T3 20040701 (200444) ES
<--
EP 409956 B2 20040721 (200449) EN
<--

APPLICATION DETAILS:

PATENT NO	KIND	APPLICATION	DATE

APPLICATION DETAILS:

PATENT NO	KIND	APPLICATION	DATE
US 5258907 A CIP of 19881221		US 1989-287481	
US 5324668 A CIP of 19890117		US 1989-297481	
US 5258907 A CIP of 19890217		US 1989-311808	
US 5324668 A CIP of 19890217		US 1989-311808	
US 5258907 A CIP of 19890508		US 1989-349373	
US 5324668 A CIP of 19890508		US 1989-349373	
US 5258907 A CIP of 19890601		US 1989-360603	
US 5324668 A CIP of 19890601		US 1989-360603	
US 5258907 A Div Ex 19891012		US 1989-420775	
US 5324668 A Cont of 19891012		US 1989-420775	
DE 69026153 E 19900116		DE 1990-69026153	
DE 69034111 E 19900116		DE 1990-69034111	
EP 409956 A 19900116		EP 1990-903086	
EP 409956 B1 19900116		EP 1990-903086	
DE 69026153 E 19900116		EP 1990-903086	
ES 2084689 T3 19900116		EP 1990-903086	
EP 666477 B1 Div Ex 19900116		EP 1990-903086	
EP 409956 B2 19900116		EP 1990-903086	
JP 03505128 W 19900116		JP 1990-503251	
JP 2644373 B2 19900116		JP 1990-503251	
EP 409956 B1		WO 1990-US291 19900116	
DE 69026153 E		WO 1990-US291 19900116	

JP 2644373 B2	WO 1990-US291 19900116
EP 409956 B2	WO 1990-US291 19900116
US 5258907 A 19910531	US 1991-709019
US 5324668 A	US 1993-51761 19930203
EP 666477 A1 19900116	EP 1995-104733
EP 666477 B1 19900116	EP 1995-104733
DE 69034111 E 19900116	EP 1995-104733
ES 2210266 T3 19900116	EP 1995-104733
EP 409956 B2 Related to 19900116	EP 1995-104733

FILING DETAILS:

PATENT NO	KIND	PATENT NO
DE 69026153 E	Based on	EP 409956 A
ES 2084689 T3	Based on	EP 409956 A
EP 666477 B1	Div ex	EP 409956 A
DE 69034111 E	Based on	EP 666477 A
ES 2210266 T3	Based on	EP 666477 A
EP 409956 B2	Related to	EP 666477 A
JP 2644373 B2	Previous Publ	JP 03505128 W
US 5258907 A	CIP of	US 5026889 A
EP 409956 B1	Based on	WO 9008325 A
DE 69026153 E	Based on	WO 9008325 A
JP 2644373 B2	Based on	WO 9008325 A
EP 409956 B2	Based on	WO 9008325 A

PRIORITY APPLN. INFO: US 1989-420775 19891012
US 1989-297481 19890117
US 1989-311808 19890217
US 1989-349373 19890508
US 1989-360603 19890602
US 1989-287481 19881221
US 1989-360603 19890601
US 1991-709019 19910531
US 1993-51761 19930203

INT. PATENT CLASSIF.:
MAIN: G01N033-76
SECONDARY: G01N033-68; G06F019-00
IPC RECLASSIF.: G01N0033-50 [I,A]; G01N0033-50 [I,A]; G01N0033-50 [I,C]; G01N0033-50 [I,C]; G01N0033-53 [I,A]; G01N0033-53 [I,C]; G01N0033-74 [I,C]; G01N0033-76 [I,A]
ECLA: G01N0033-76
USCLASS NCLM: 436/510.000
NCLS: 435/007.900; 435/007.920; 436/086.000; 436/087.000; 436/510.000; 436/817.000; 436/818.000

BASIC ABSTRACT:
WO 1990008325 A UPAB: 20050630 (A) A method for determining if a pregnant worman is at significant risk of carrying a fetus with down syndrome (DS) is claimed comprising measuring a pregnant women's material serum level of free beta subunit of human chorionic gonadotropin (hCG), incorporating the measurement of the level and the pregnant womens gestational age into a probability density function to compare with a set of normative data to

determine the pregnant womans risk of carrying a fetus with DS. (B) Also claimed is a method for determining if a pregnant women is at significant risk of carrying a fetus with DS comprising assaying a pregnant womens blood for free beta subunit of HCG, the results of the assay being indicative of increased risk of fetal DS. The method may further comprise assaying a pregnant womens blood for alpha-fetoprotein (A), (C) Also claimed is an assay for measuring a persons blood level of the free beta subunit of hCG, (D) Also claimed is an appts. for receiving a measurement of a pregnant womens maternal blood level of the free beta subunit of hCG and a computer for comparing the measurement of the level to a set of reference data to determine fetal chromosomal abnormalities.

ADVANTAGE - The method correctly predicts a higher percentage of fetal DS cases with a lesser false positive rate than other known methods. Detection efficiency for Trisomy 21 as high as 83% has been achieved. The method can also be used for detecting chromosomal trisomies such as trisomy 13 and trisomy 18.

FILE SEGMENT: CPI; EPI
MANUAL CODE: CPI: B04-B02D4; B04-B04D5; B04-B04L; B11-C; B11-C08; B12-K04A3; D05-H09
EPI: S03-E14H; S05-C

=> fil hcap
FILE 'HCAPLUS' ENTERED AT 16:32:25 ON 15 DEC 2008
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FILE COVERS 1907 - 15 Dec 2008 VOL 149 ISS 25
FILE LAST UPDATED: 14 Dec 2008 (20081214/ED)

HCAplus now includes complete International Patent Classification (IPC) reclassification data for the third quarter of 2008.

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=> fil biosis
FILE 'BIOSIS' ENTERED AT 16:32:29 ON 15 DEC 2008

FILE COVERS 1926 TO DATE.
CAS REGISTRY NUMBERS AND CHEMICAL NAMES (CNs) PRESENT FROM JANUARY 1926 TO DATE.

RECORDS LAST ADDED: 10 December 2008 (20081210/ED)

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BIOSIS has been augmented with 1.8 million archival records from 1926
through 1968.  These records have been re-indexed to match current
BIOSIS indexing.

=> fil embase
FILE 'EMBASE' ENTERED AT 16:32:33 ON 15 DEC 2008
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 FILE COVERS 1974 TO 15 Dec 2008 (20081215/ED)

 EMBASE was reloaded on March 30, 2008.

 EMBASE is now updated daily.  SDI frequency remains weekly (default)
 and biweekly.

 This file contains CAS Registry Numbers for easy and accurate
 substance identification.

 Beginning January 2008, Elsevier will no longer provide EMTREE
 codes as part of the EMTREE thesaurus in EMBASE.  Please update
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 codes.

 For further assistance, please contact your local helpdesk.

=> fil medline
FILE 'MEDLINE' ENTERED AT 16:32:37 ON 15 DEC 2008

 FILE LAST UPDATED: 11 Dec 2008 (20081211/UP).  FILE COVERS 1949 TO DATE.

  MEDLINE has been updated with the National Library of Medicine's
  revised 2008 MeSH terms.  See HELP RLOAD for details.

  This file contains CAS Registry Numbers for easy and accurate
  substance identification.

  See HELP RANGE before carrying out any RANGE search.

  MEDLINE Accession Numbers (ANs) for records from 1950-1977 have
  been converted from 8 to 10 digits.  Searches using an 8 or 10 digit
  AN will retrieve the same record.  The 10-digit ANs can be expanded,
  searched, and displayed in all records from 1949 to the present.

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L7          508 SEA FILE=HCAPLUS ABB=ON  PLU=ON  L6 AND (L4 OR L5)
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L9              QUE  ABB=ON  PLU=ON  SCREEN?
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L70 ANSWER 1 OF 25 HCAPLUS COPYRIGHT 2008 ACS on STN
ACCESSION NUMBER: 2007:1454254 HCAPLUS Full-text
DOCUMENT NUMBER: 148:96046
ENTRY DATE: Entered STN: 24 Dec 2007
TITLE: Diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection
INVENTOR(S): Kapur, Ravi; Toner, Mehmet; Wang, Zihua; Fuchs, Martin
PATENT ASSIGNEE(S): Living Microsystems, Inc., USA; CellPoint Diagnostics, Inc.; The General Hospital Corporation
SOURCE: PCT Int. Appl., 92 pp.
CODEN: PIXXD2
DOCUMENT TYPE: Patent
LANGUAGE: English
CLASSIFICATION: 9-11 (Biochemical Methods)
Section cross-reference(s): 3, 14
FAMILY ACC. NUM. COUNT: 7
PATENT INFORMATION:

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
WO 2007147076	A2	20071221	WO 2007-US71250	200706 14
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WO 2007147076	A3	20080403		
W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BH, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DO, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IS, JP, KE, KG, KM, KN, KP, KR, KZ, LA, LC, LK, LR, LS, LT, LU, LY, MA, MD, ME, MG, MK, MN, MW, MX, MY, MZ, NA, NG, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RS, RU, SC, SD, SE, SG, SK, SL, SM, SV, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, ZA, ZM, ZW				
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US 20080050739	A1	20080228	US 2007-763426	200706 14
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US 20080138809	A1	20080612	US 2007-763245	200706 14
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PRIORITY APPLN. INFO.: US 2006-804817P P 200606

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US 2006-820778P P
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US 2006-804815P P
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US 2006-804818P P
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PATENT CLASSIFICATION CODES:

PATENT NO.	CLASS	PATENT FAMILY CLASSIFICATION CODES
WO 2007147076	IPCI	C12Q0001-68 [I,A]; C12Q0001-68 [I,C]; C12Q0001-68 [I,A]; C12P0019-00 [I,C]; C12P0019-34 [I,A]; G01N0033-48 [I,C]; G01N0033-48 [I,A]
	IPCR	C12Q0001-68 [I,C]; C12Q0001-68 [I,A]; C12P0019-00 [I,C]; C12P0019-34 [I,A]; G01N0033-48 [I,C]; G01N0033-48 [I,A]
	ECLA	C12Q001/68A4
US 20080026390	IPCI	C12Q0001-68 [I,A]
	NCL	435/006.000
	ECLA	C12Q001/68M6; C12Q001/68A6; C12Q001/68B6
US 20080050739	IPCI	C12Q0001-68 [I,A]; G06G0007-48 [I,A]; G06G0007-00 [I,C*]
	NCL	435/006.000; 703/011.000
US 20080138809	IPCI	C12Q0001-68 [I,A]; C12Q0001-02 [I,A]

ABSTRACT:
The present invention relates to methods for detecting, enriching, and analyzing rare cells that are present in the blood, e.g. fetal cells. The method includes the prenatal detection of ***chromosomal*** abnormalities, genetic polymorphisms ***detection*** , and cancer risk assessment. The invention further features automated methods of analyzing rare cell(s) to determine the presence of an abnormality, disease or condition in a subject, e.g. a ***fetus*** by analyzing a cellular sample from the subject. Thus, microfluidic devices of the invention were designed by computer -aided design (CAD) and microfabricated by photolithog. A two-step process was developed in which a blood sample is first debulked to remove the large population of small cells, and then the rare target epithelial cells target cells are recovered by immunoaffinity capture.

SUPPL. TERM: prenatal diagnosis fetal cell enriched sample maternal blood; fetus chromosome abnormality detection cord blood enriched sample; SNP detection genetic disease susceptibility cancer diagnosis enriched sample

INDEX TERM: Computers
(-aided design (CAD), of microfluidic devices; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Nervous system, disease
(Charcot-Marie-Tooth; diagnostic methods

using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Bone, neoplasm
(Ewing's sarcoma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Sarcoma
(Ewing's; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Neoplasm
(Giant cell; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Sarcoma
(Kaposi's; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Testis, disease
(Klinefelter's syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Trisomy
(Patau's syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Transcription factors
ROLE: BSU (Biological study, unclassified); BIOL (Biological study)
(TDF (testis-determining factor), gene on Y, syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Sarcoma
(Veticulum cell; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Chromosome disorders
(Williams syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Kidney, neoplasm
(Wilms'; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Lymphocyte
Polymorphonuclear leukocyte
(acute or chronic lymphocyctic or granulocytic

tumor; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Amniotic fluid
Cord blood
Endothelium
Epithelium
Pregnancy
Stem cell
(anal.; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Oligonucleotides
ROLE: ARG (Analytical reagent use); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(analogs; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Fertility disorders
(azoospermia; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Skin, neoplasm
(basal cell carcinoma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Carcinoma
(basal cell; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Spheres
(beads, amplifying occurs on bead; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Diagnosis
(cancer; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Skin, neoplasm
(carcinoma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Tumor markers
(cell separation using; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Microarray technology
(cell size-based separation using; diagnostic

methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Separation
(cell size-based; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Uterus, disease
(cervix, dysplasia; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Intestine, neoplasm
(colon; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Adrenal cortex, disease
(congenital adrenal hypoplasia; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Chromosome disorders
(crying cat syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Carcinoma
(cutaneous squamous cell; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Carcinoma
(cutaneous; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Polymerase chain reaction
(degenerate oligonucleotide primed; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Mutation
(deletion, chromosomal, detecting; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Alleles
Chromosome aberrations
Single nucleotide polymorphism
(detecting; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Acute lymphocytic leukemia
Acute myeloid leukemia
Acute promyelocytic leukemia
Adenocarcinoma
Adenoma
Adrenal gland, neoplasm
Aneuploidy
Blood analysis
Bone, neoplasm
Brain, neoplasm
Bronchi, neoplasm
Carcinoid
Chronic myeloid leukemia
DiGeorge syndrome
Down's syndrome
Fetus
Flow cytometry
Gallbladder, neoplasm
Head and Neck, neoplasm
Human
Hyperplasia
Kallmann syndrome
Kidney, neoplasm
Larynx, neoplasm
Liver, neoplasm
Lung, neoplasm
Lymphoma
Mammary gland, neoplasm
Melanoma
Microfluidic devices
Multiple myeloma
Mycosis fungoides
Myelodysplastic syndromes
Nerve, neoplasm
Neurofibromatosis
Ovary, neoplasm
Pancreas, neoplasm
Parathyroid gland, neoplasm
Pelizaeus-Merzbacher disease
Pheochromocytoma
Polycythemia vera
Preeclampsia
Prostate gland, neoplasm
Quality control
Skin, neoplasm
Small-cell lung carcinoma
Stomach, neoplasm
Susceptibility (genetic)
Test kits
Thyroid gland, neoplasm
(diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: RNA
ROLE: ANT (Analyte); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(diagnostic methods using rare cell-enriched samples, particularly, in prenatal or

cancer diagnosis, and polymorphisms detection)

INDEX TERM: Primers (nucleic acid)
Probes (nucleic acid)
ROLE: ARG (Analytical reagent use); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Mutation
(duplication, chromosomal, detection; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Uterus, disease
(endometriosis; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Nucleic acids
ROLE: ANT (Analyte); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(fetal; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Neoplasm
(gallstone; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Nerve, neoplasm
(ganglioneuroma, Intestinal; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Risk assessment
(genetic disease; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Disease, animal
(genetic, Alagille syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Disease, animal
(genetic, Cat eye syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Disease, animal
(genetic, Smith-Magenis syndrome;

diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Disease, animal
(genetic, Wolf-Hirschhorn syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Diagnosis
(genetic; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: DNA
ROLE: ANT (Analyte); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(genomic; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Neuroglia, neoplasm
(glioblastoma, multiforma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Neoplasm
(hairy-cell; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Neoplasm
(head and neck; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: DNA sequence analysis
(high throughput, in diagnosis; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Chromosome
(human 1, 1p26 deletion, syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Chromosome
(human 13, anal.; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Trisomy
(human 13; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms

detection)
INDEX TERM: Chromosome
(human 17, dup(17)(p11.2p11.2), syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)
INDEX TERM: Chromosome
(human 18, anal.; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)
INDEX TERM: Trisomy
(human 18; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)
INDEX TERM: Chromosome
(human 21, anal.; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)
INDEX TERM: Chromosome
(human 22, dup(22)(q11.2q11.2), syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)
INDEX TERM: Chromosome
(human X, anal.; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)
INDEX TERM: Chromosome
(human Y, abnormality, detection ; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)
INDEX TERM: Neoplasm
(humoral hypercalcemia of malignancy; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)
INDEX TERM: Pressure
(hyperbaric or hypobaric, cell separation using; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)
INDEX TERM: Neoplasm
(hyperplastic corneal nerve; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)
INDEX TERM: Oligonucleotides
ROLE: ARG (Analytical reagent use); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL

(Biological study); USES (Uses)
(immobilized; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: DNA microarray technology
Genotyping (method)
Nucleic acid amplification
Raman spectroscopy
(in diagnosis; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Carcinoma
(in situ; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Oligonucleotides
ROLE: ARG (Analytical reagent use); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(labeled; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Myoma
(leiomyoma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Blood
(maternal, fetal-cell-enriched sample from; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Photolithography
(microfluidic devices microfabricated by; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Eye, disease
Skin, disease
(microphthalmia/linear skin defect; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Genetic polymorphism
(microsatellite, STR, detecting; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Nucleic acid amplification
(multiple displacement amplification; diagnostic methods using rare cell-enriched

samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Nerve, neoplasm
(neuroblastoma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Nerve, neoplasm
(neuroma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Nerve, disease
(neuropathy, with liability to pressure palsies; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Chemiluminescent substances
Chromophores
Dyes
Fluorescent substances
Magnetic materials
Phosphorescent substances
Radioactive substances
(oligonucleotides labeled with; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Antigens
Enzymes, biological studies
Heavy metals
ROLE: ARG (Analytical reagent use); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(oligonucleotides labeled with; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Bone, neoplasm
Sarcoma
(osteosarcoma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Antibodies and Immunoglobulins
Carbohydrates, biological studies
Ligands
Nucleic acids
Proteins
Receptors
ROLE: ARG (Analytical reagent use); BSU (Biological study, unclassified); TEM (Technical or engineered material use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(particular cell-binding, cell separation using; diagnostic methods using rare cell-enriched

samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Parturition disorders
(premature parturition; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Diagnosis
(prenatal; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Nucleic acid amplification
(primer extension pre-amplification, and improved primer extension pre-amplification; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Sample preparation
(rare cell-enriched; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Biomarkers
(rare cells selected using; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Polymerase chain reaction
(real-time, in diagnosis; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Intestine, neoplasm
(rectum; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Eye, neoplasm
(retinoblastoma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Sarcoma
(rhabdomyosarcoma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Nanoparticles
(scattering or fluorescent, oligonucleotides labeled with; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Testis, neoplasm

(seminoma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Synthesis
(sequencing by, in diagnosis; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Sarcoma
(soft-tissue; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Skin, neoplasm
(squamous cell carcinoma; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Magnetic field
(that selectively retain paramagnetic components, cell separation using; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Pancreatic islet of Langerhans
(tumor; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: Disease, animal
(velo-cardio-facial syndrome; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: 9025-62-1, Steroid sulfatase 9030-66-4, Glycerol kinase
ROLE: BSU (Biological study, unclassified); BIOL (Biological study)
(deficiency; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

INDEX TERM: 1000082-89-2 1000082-90-5 1000082-91-6
1000082-92-7 1000082-93-8 1000082-94-9
1000082-95-0 1000082-96-1 1000082-97-2
1000082-98-3 1000082-99-4 1000083-00-0
1000083-01-1 1000083-02-2 1000083-03-3
1000083-04-4 1000083-05-5 1000083-06-6
1000083-07-7 1000083-08-8 1000083-09-9
1000083-10-2 1000083-11-3 1000083-12-4
1000083-13-5 1000083-14-6 1000083-15-7
1000083-16-8 1000083-17-9 1000083-18-0
1000083-19-1 1000083-20-4 1000083-21-5
1000083-22-6 1000083-23-7 1000083-24-8
1000083-25-9 1000083-26-0 1000083-27-1
1000083-28-2 1000083-29-3 1000083-30-6

1000083-31-7 1000083-32-8 1000083-33-9
1000083-34-0 1000083-35-1 1000083-36-2
1000083-37-3 1000083-38-4 1000083-39-5
1000083-40-8 1000083-41-9 1000083-42-0
1000083-43-1 1000083-44-2 1000083-45-3
1000083-46-4 1000083-47-5 1000083-48-6
1000083-49-7 1000083-50-0 1000083-51-1
1000083-52-2 1000083-53-3 1000083-54-4
1000083-55-5 1000083-56-6 1000083-57-7
1000083-58-8 1000083-59-9 1000083-60-2
1000083-61-3 1000083-62-4 1000083-63-5
1000083-64-6 1000083-65-7 1000083-66-8
1000083-67-9 1000083-68-0 1000083-69-1
1000083-70-4 1000083-71-5 1000083-72-6
1000083-73-7 1000083-74-8 1000083-75-9
1000083-76-0 1000083-77-1 1000083-78-2
1000083-79-3 1000083-80-6 1000083-81-7
1000083-82-8 1000083-83-9 1000083-84-0
1000083-85-1 1000083-86-2 1000083-87-3
1000083-88-4 1000083-89-5 1000083-90-8
1000083-91-9 1000083-92-0 1000083-93-1
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1000083-97-5 1000083-98-6 1000083-99-7
1000084-00-3 1000084-01-4 1000084-02-5
1000084-03-6 1000084-04-7 1000084-05-8
1000084-06-9 1000084-07-0 1000084-08-1
1000084-09-2 1000084-10-5 1000084-11-6
1000084-12-7 1000084-13-8 1000084-14-9
1000084-15-0 1000084-16-1 1000084-17-2
1000084-18-3 1000084-19-4 1000084-20-7
1000084-21-8 1000084-22-9 1000084-23-0
1000084-24-1 1000084-25-2 1000084-26-3
1000084-27-4 1000084-28-5 1000084-29-6
1000084-30-9 1000084-31-0 1000084-32-1
1000084-33-2 1000084-34-3 1000084-35-4
1000084-36-5 1000084-37-6 1000109-37-4
1000109-38-5 1000109-39-6 1000109-40-9
1000109-41-0 1000109-42-1 1000109-43-2
1000109-44-3 1000109-45-4 1000109-46-5
ROLE: PRP (Properties)
(unclaimed nucleotide sequence; diagnostic methods using rare cell-enriched samples, particularly, in prenatal or cancer diagnosis, and polymorphisms detection)

L70 ANSWER 2 OF 25 HCAPLUS COPYRIGHT 2008 ACS on STN
ACCESSION NUMBER: 2007:1116340 HCAPLUS Full-text
DOCUMENT NUMBER: 147:403737
ENTRY DATE: Entered STN: 04 Oct 2007
TITLE: Screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood
INVENTOR(S): Bischoff, Farideh Z.; Simpson, Joe Leigh
PATENT ASSIGNEE(S): Baylor College of Medicine, USA
SOURCE: PCT Int. Appl., 29pp.
CODEN: PIXXD2
DOCUMENT TYPE: Patent
LANGUAGE: English
CLASSIFICATION: 14-14 (Mammalian Pathological Biochemistry)

Section cross-reference(s): 3, 9
FAMILY ACC. NUM. COUNT: 1
PATENT INFORMATION:

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
WO 2007112418	A2	20071004	WO 2007-US65295	200703 27
			<--	
WO 2007112418	A3	20081023		
W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BH, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IS, JP, KE, KG, KM, KN, KP, KR, KZ, LA, LC, LK, LR, LS, LT, LU, LY, MA, MD, MG, MK, MN, MW, MX, MY, MZ, NA, NG, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RS, RU, SC, SD, SE, SG, SK, SL, SM, SV, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, ZA, ZM, ZW				
RW: AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IS, IT, LT, LU, LV, MC, MT, NL, PL, PT, RO, SE, SI, SK, TR, BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG, BW, GH, GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM, AP, EA, EP, OA				
US 20080038733	A1	20080214	US 2007-692115	200703 27
			<--	
PRIORITY APPLN. INFO.:			US 2006-786660P P	200603 28
			<--	
			US 2007-692115 A	200703 27

PATENT CLASSIFICATION CODES:

PATENT NO.	CLASS	PATENT FAMILY CLASSIFICATION CODES
WO 2007112418	IPCI	C12Q0001-68 [I,A]; C12Q0001-68 [I,C]; C12Q0001-68 [I,A]
	IPCR	C12Q0001-68 [I,C]; C12Q0001-68 [I,A]
	ECLA	C12Q001/68M6
US 20080038733	IPCI	C12Q0001-68 [I,A]
	NCL	435/006.000
	ECLA	C12Q001/68M6

ABSTRACT:
The present disclosure describes methods for screening and ***identifying*** genomic sequences useful in estimating the risk of fetal aneuploidy, particularly trisomy 21. This disclosure also describes methods for utilizing such genomic sequences alone or to augment existing non-invasive diagnostics for Trisomy 21 and other aneuploidies. Particularly, the methods based on the anal. of fetal non-Y chromosome DMA from bodily fluid of a pregnant woman, particularly, from whole blood collected from a finger prick and spoted onto standard blood specimen cards. Provided are primers and probes for fetal beta-globin genomic locus for use in real-time RT-PCR amplification anal.

SUPPL. TERM: prenatal diagnosis fetus

aneuploidy Down syndrome mother blood; fetal beta globin gene PCR primer screening mother blood

INDEX TERM: Fetus
(DNA anal.; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Extraction
(DNA, fetal, from mother's blood; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Pregnancy
(aneuploid, control data set matched to test sample for history of; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: DNA
ROLE: ANT (Analyte); DGN (Diagnostic use); PUR (Purification or recovery); ANST (Analytical study); BIOL (Biological study); PREP (Preparation); USES (Uses)
(fetal, non-Y chromosome; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Aneuploidy
(fetal; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Pregnancy
(first trimester, anal.; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Gene, animal
ROLE: ADV (Adverse effect, including toxicity); ANT (Analyte); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(for fetal β-globin; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Diagnosis
(genetic, non-invasive; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Pregnancy
(gestational age, control data set matched to test sample for; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Aging, animal
(maternal age, control data set matched to test sample for; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Diabetes mellitus
(maternal diabetic status, control data set matched to test sample for; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Human groups
(maternal race status, control data set matched to

test sample for; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Body weight
(maternal, control data set matched to test sample for; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Probability
(of aneuploidy, Multiplicity of Median value exceeds threshold empirically determined to correspond to; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Body fluid
(of pregnant woman, anal.; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Diagnosis
(prenatal; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Polymerase chain reaction
(real-time; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Data processing
Down's syndrome
Human
Prognosis
Risk assessment
Statistical analysis
Susceptibility (genetic)
(screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Behavior
(smoking, maternal status, control data set matched to test sample for; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Primers (nucleic acid)
ROLE: ARG (Analytical reagent use); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(specific for fetal beta-globin genomic locus; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Regression analysis
(weighted log-linear; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Blood analysis
(whole; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: Hemoglobins
ROLE: BSU (Biological study, unclassified); BIOL (Biological study)

(β chain, fetal, genomic locus for, anal.; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: 951182-01-7 951182-02-8 951182-03-9 951182-04-0 951182-05-1 951182-06-2
ROLE: ARG (Analytical reagent use); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(control primer; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: 951182-07-3
ROLE: ARG (Analytical reagent use); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(primer specific for fetal beta-globin genomic locus; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: 951182-08-4
ROLE: ARG (Analytical reagent use); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(primer, specific for fetal beta-globin genomic locus; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

INDEX TERM: 951182-09-5
ROLE: ARG (Analytical reagent use); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(probe, specific for fetal beta-globin genomic locus; screening for fetal aneuploidy, particularly Down syndrome, using fetal DNA isolated from mother's blood)

L70 ANSWER 3 OF 25 MEDLINE on STN
ACCESSION NUMBER: 2005677933 MEDLINE Full-text
DOCUMENT NUMBER: PubMed ID: 16353275
TITLE: The effect of fetal gender on the false-positive rate of Down syndrome by maternal serum screening.
AUTHOR: Mueller V M; Huang T; Summers A M; Winsor S H M
CORPORATE SOURCE: Division of Maternal Fetal Medicine, Department of Obstetrics and Gynecology, McMaster University, Hamilton, Ontario, Canada.. muellevm@mcmaster.ca
SOURCE: Prenatal diagnosis, (2005 Dec) Vol. 25, No. 13, pp. 1258-61.
Journal code: 8106540. ISSN: 0197-3851.
PUB. COUNTRY: England: United Kingdom
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200704
ENTRY DATE: Entered STN: 22 Dec 2005
Last Updated on STN: 12 Dec 2006
Entered Medline: 12 Apr 2007
ABSTRACT:
OBJECTIVES: (1) To further explore if there is a difference in maternal

serum levels of alpha-fetoprotein (AFP), human chorionic gonadotrophin (hCG) and estriol (uE3) between fetal genders. (2) To determine if these differences influence false-positive rates of Down ***syndrome*** screening in pregnancies with male or female fetuses. METHODS: This is a descriptive study of women screened at the Ontario Maternal Serum Screening program between 1993 and 1995. The women were grouped by fetal gender and ethnicity. Serum levels of the three markers and screening false-positive rates for Down syndrome were compared between fetal genders in women of different ethnicity respectively. RESULTS: Complete data were available for 110 306 pregnancies. In all three ethnic groups, MSAFP levels were significantly decreased and MShCG levels were significantly increased in women with female fetuses. The level of MSuE3 was similar between genders. The difference in false-positive rates of Down syndrome between genders was not ***statistically*** significant. CONCLUSIONS: This is the largest study comparing false-positive rates between fetal genders. In contrast to previous studies, the differences in the serum marker levels between fetal genders do not influence the false-positive rates for Down syndrome.

CONTROLLED TERM: Check Tags: Female; Male
Adult
African Continental Ancestry Group
Asian Continental Ancestry Group
*Chorionic Gonadotropin: BL, blood
Down Syndrome: BL, blood
*Down Syndrome: DI, diagnosis
Down Syndrome: EH, ethnology
*Estriol: BL, blood
European Continental Ancestry Group
False Positive Reactions
Gestational Age
Humans
Mass Screening: MT, methods
Ontario: EP, epidemiology
Pregnancy
*Sex Characteristics
*alpha-Fetoproteins: AN, analysis

CAS REGISTRY NO.: 50-27-1 (Estriol)

CHEMICAL NAME: 0 (Chorionic Gonadotropin); 0 (alpha-Fetoproteins)

L70 ANSWER 4 OF 25 EMBASE

ACCESSION NUMBER: 2005367836 EMBASE Full-text

TITLE: SURUSS in perspective.

AUTHOR: Wald, N.J. (correspondence); Hackshaw, A.K.; Rudnicka, A.

CORPORATE SOURCE: Department of Environmental and Preventive Medicine, Wolfson Institute of Preventive Medicine, Barts and the London School of Medicine and Dentistry, United Kingdom. n.j.wald@qmul.ac.uk

AUTHOR: Rodeck, C.

CORPORATE SOURCE: Department of Obstetrics and Gynaecology, University College London, United Kingdom.

AUTHOR: Wald, N.J. (correspondence)

CORPORATE SOURCE: Wolfson Institute of Preventive Medicine, Barts and the London School of Medicine and Dentistry, Charter house Square, London EC1M 6BQ, United Kingdom. n.j.wald@qmul.ac.uk

SOURCE: Seminars in Perinatology, (Aug 2005) Vol. 29, No. 4, pp. 225-235.
Refs: 27
ISSN: 0146-0005 CODEN: SEMPDU
PUBLISHER IDENT.: S 0146-0005(05)00040-6
COUNTRY: United States
DOCUMENT TYPE: Journal; Conference Article; (Conference paper)
FILE SEGMENT: 010 Obstetrics and Gynecology
014 Radiology
022 Human Genetics
029 Clinical and Experimental Biochemistry
007 Pediatrics and Pediatric Surgery
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 15 Sep 2005
Last Updated on STN: 15 Sep 2005

ABSTRACT: BACKGROUND: Until the publication of the Serum Urine and Ultrasound Screening Study (SURUSS) report, it was difficult to compare the different antenatal screening tests for Down's ***Syndrome*** because of variations in study designs. We here present the main results from SURUSS, updated to take account of recent information on nuchal translucency in Down's Syndrome ***pregnancies*** , and discuss their implications. METHODS: SURUSS was a prospective study of 47,053 singleton pregnancies (including 101 ***pregnancies*** with Down's Syndrome) conducted in 25 maternity units. Nuchal translucency measurements were taken. Serum and urine samples collected between 9 and 13 weeks, and again between 14 and 20 weeks of pregnancy were stored. Samples from each affected pregnancy and five matched controls were tested for currently used or suggested biochemical Down's Syndrome ***screening*** markers. Pregnancies were followed up to determine the presence or absence of Down's Syndrome. For an 85% ***Down*** 's Syndrome detection rate, the false-positive rate for the Integrated test (nuchal translucency and pregnancy associated plasma protein-A [PAPP-A] at 11 completed weeks of pregnancy, and α-fetoprotein, unconjugated oestriol [uE(3)], free β or total human chorionic gondaotrophin (hCG) and inhibin-A in the early second trimester) was 0.9%, the Serum integrated test (without nuchal translucency) 2.7%, the Combined test (nuchal translucency with free β-hCG and PAPP-A at 11 weeks) 4.3%, the Quadruple test (α-fetoprotein, uE (3), free β or total hCG and inhibin-A) 6.2%, and nuchal translucency at 11 weeks, 15.2%. All tests included maternal age. Using the Integrated test at an 85% detection rate, there would be six diagnostic procedure-related unaffected fetal losses following amniocentesis per 100,000 women screened compared with 35 using the Combined test or 45 with the Quadruple test. CONCLUSIONS: The Integrated test offers the most effective and safe method of screening for women who attend in the first trimester. The next best test is the Serum integrated test. The Quadruple test is the best test for women who first attend in the second trimester. There is no justification for retaining the Double (α-fetoprotein and hCG) or Triple (α-fetoprotein, uE(3), and hCG) tests, or nuchal translucency alone (with or without maternal age) in antenatal screening for Down's Syndrome.

CONTROLLED TERM: Medical Descriptors:
adult
amniocentesis

blood analysis
comparative study
conference paper
controlled study
diagnostic procedure
diagnostic test
*Down syndrome: CN, congenital disorder
*Down syndrome: DI, diagnosis
female
*fetus echography
first trimester pregnancy
follow up
gestational age
human
major clinical study
maternal age
maternal serum
maternity ward
pregnancy
prenatal period
*prenatal screening
priority journal
screening test
second trimester pregnancy
statistical analysis
urinalysis

CONTROLLED TERM: Drug Descriptors:
alpha fetoprotein: EC, endogenous compound
biochemical marker: EC, endogenous compound
chorionic gonadotropin: EC, endogenous compound
estriol: EC, endogenous compound
inhibin A: EC, endogenous compound
pregnancy associated plasma protein A: EC, endogenous compound

CAS REGISTRY NO.: (chorionic gonadotropin) 9002-61-3; (estriol) 50-27-1

L70 ANSWER 5 OF 25 EMBASE COPYRIGHT (c) 2008 Elsevier B.V. All rights reserved on STN

ACCESSION NUMBER: 2004363737 EMBASE Full-text

TITLE: Clinical application of inhibin A measurement: Prenatal serum screening for Down syndrome.

AUTHOR: Lambert-Messerlian, Geralyn M., Dr. (correspondence)

CORPORATE SOURCE: Prenatal and Special Testing, Women and Infants Hospital, 70 Elm Street, Providence, RI 02903, United States.

AUTHOR: Lambert-Messerlian, Geralyn M., Dr. (correspondence); Canick, Jacob A.

CORPORATE SOURCE: Dept. of Pathol. and Lab. Medicine, Div. of Prenatal and Special Testing, Women/Infants Hosp./Brown Med. Sch., Providence, RI, United States.

SOURCE: Seminars in Reproductive Medicine, (Aug 2004) Vol. 22, No. 3, pp. 235-242.
Refs: 73
ISSN: 1526-8004 CODEN: SRMECJ

COUNTRY: United States

DOCUMENT TYPE: Journal; General Review; (Review)

FILE SEGMENT: 010 Obstetrics and Gynecology
003 Endocrinology
007 Pediatrics and Pediatric Surgery

LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 16 Sep 2004
Last Updated on STN: 16 Sep 2004

ABSTRACT: Inhibin A is secreted in significant quantities by the corpus luteum and fetoplacental unit, suggesting a role in fertility and pregnancy. Negative feedback regulation of follicle-stimulating hormone during pregnancy is one expected function of inhibin A, but the complete repertoire of actions of this hormone in pregnancy, including paracrine and autocrine actions, is yet to be fully understood. Inhibin A levels have been carefully described throughout normal pregnancy and studied in association with maternal and fetal complication such as intrauterine growth restriction, preterm labor or delivery, and preeclampsia. The ***first*** clinical application of inhibin A measurement in pregnancy has been its use as a second-trimester maternal serum ***marker*** for Down syndrome. Our laboratory was among the ***first*** , in 1998, to implement Quad marker ***screening*** , inhibin A measurement in conjunction with α-fetoprotein, unconjugated estriol, and human chorionic gonadotropin, to assess patients' risk of having a Down syndrome baby. The test performance of the Quad test has been validated by several large studies, detecting about 80% of Down syndrome ***pregnancies*** at a 5% false-positive rate. The present review describes Down syndrome and the use of inhibin A in second -trimester prenatal screening. We also address the method used for inhibin A measurement, the biology of inhibin A in Down ***syndrome*** pregnancy, and the effects of covariates and other fetal abnormalities on inhibin A levels.

CONTROLLED TERM: Medical Descriptors:
corpus luteum function
covariance
*Down syndrome: CN, congenital disorder
*Down syndrome: DI, diagnosis
female
fertility
fetoplacental unit
fetus
fetus disease
human
intrauterine growth retardation: CO, complication
laboratory diagnosis
negative feedback
preeclampsia
premature labor
*prenatal diagnosis
quad marker screening
quantitative diagnosis
review
screening
second trimester pregnancy
statistical significance

CONTROLLED TERM: Drug Descriptors:
alpha fetoprotein: EC, endogenous compound
chorionic gonadotropin: EC, endogenous compound
estriol: EC, endogenous compound
follitropin: EC, endogenous compound
*inhibin A: EC, endogenous compound

CAS REGISTRY NO.: (chorionic gonadotropin) 9002-61-3; (estriol) 50-27-1; (follitropin) 9002-68-0

L70 ANSWER 6 OF 25 BIOSIS COPYRIGHT (c) 2008 The Thomson Corporation on STN DUPLICATE 1
ACCESSION NUMBER: 2003:38231 BIOSIS Full-text
DOCUMENT NUMBER: PREV200300038231
TITLE: Early genetic sonogram for Down syndrome detection.
AUTHOR(S): Bahado-Singh, Ray O. [Reprint Author]; Mendilcioglu, Inanc; Rowther, Minu; Choi, Sang-Joon; Oz, Utku; Yousefi, Nastaran Foyouzi; Mahoney, Maurice J.
CORPORATE SOURCE: Department of Obstetrics and Gynecology, University of Cincinnati, 231 Albert Sabin Way, MI, 0526, Cincinnati, OH, 45267-0526, USA
bahadoro@ucmail.uc.edu
SOURCE: American Journal of Obstetrics and Gynecology, (November 2002) Vol. 187, No. 5, pp. 1235-1238. print.
CODEN: AJOGAH. ISSN: 0002-9378.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 8 Jan 2003
Last Updated on STN: 8 Jan 2003
ABSTRACT:OBJECTIVE: The purpose of this study was to determine the Down syndrome sensitivity of early genetic sonography (14-<16 weeks of gestation) and to compare its diagnostic accuracy with that later in the mid trimester (16-24 weeks of gestation). STUDY DESIGN: Nuchal thickness, humerus and femur lengths, hyperechoic bowel, hypoplastic fifth digit (clinodactyly), and any gross anatomic defects were measured or ascertained in singleton pregnancies that were undergoing genetic amniocentesis. Multiple stepwise logistic regression analysis was used to determine the significant sonographic ***markers*** for Down syndrome detection in each group. Multivariate gaussian algorithms that included maternal age were used to estimate patient-specific Down syndrome risk. Sensitivity and false-positive rates, receiver-operating characteristic curves, and area under the curves were calculated and compared for both groups. RESULTS: There were 1727 pregnancies with 22 ***Down*** syndrome fetuses (1.27%) in the early group versus 3914 pregnancies with 86 Down ***syndrome*** fetuses (2.2%) in the later group. The mean +- SD ages were 15.5+-0.4 weeks versus 17.6+-1.4 weeks, respectively. Early genetic sonography (14-<16 weeks) had a 100% detection rate, with a 21.2% false-positive rate. The early versus later genetic sonography had an 81.8% versus 61.6% detection rate, respectively, at a fixed 4.8% false-positive rate. Early sonography had significantly higher diagnostic accuracy (area under the curve, 0.962 vs 0.871, respectively; P=.005). In fetuses at 14 to 15 weeks, the genetic sonography was also highly accurate, with 100% detection with a 21.9% false-positive rate. CONCLUSION: Early genetic sonography is highly sensitive and ***statistically*** superior to later ultrasonography for Down ***syndrome*** detection. Early midtrimester sonography achieved a diagnostic accuracy similar to that currently reported for first-trimester nuchal translucency.
CONCEPT CODE: Radiation biology - Radiation and isotope techniques 06504
Behavioral biology - Human behavior 07004
Pathology - Diagnostic 12504
Reproductive system - Physiology and biochemistry 16504
Reproductive system - Pathology 16506

Bones, joints, fasciae, connective and adipose tissue - Physiology and biochemistry 18004
Nervous system - Pathology 20506
Psychiatry - Psychopathology, psychodynamics and therapy 21002
Development and Embryology - General and descriptive 25502
Development and Embryology - Pathology 25503

INDEX TERMS: Major Concepts
Neurology (Human Medicine, Medical Sciences); Obstetrics (Human Medicine, Medical Sciences); Psychiatry (Human Medicine, Medical Sciences); Radiology (Medical Sciences)

INDEX TERMS: Parts, Structures, & Systems of Organisms
femur: skeletal system, length; humerus: skeletal system, length

INDEX TERMS: Diseases
Down syndrome: behavioral and mental disorders, congenital disease, nervous system disease, diagnosis
Down Syndrome (MeSH)

INDEX TERMS: Methods & Equipment
early genetic sonography: clinical techniques, diagnostic techniques; genetic amniocentesis: genetic techniques, laboratory techniques

INDEX TERMS: Miscellaneous Descriptors
diagnostic accuracy; maternal age; nuchal thickness; nuchal translucency; pregnancy

ORGANISM: Classifier
Hominidae 86215
Super Taxa
Primates; Mammalia; Vertebrata; Chordata; Animalia
Organism Name
human (common): fetus, patient, female
Taxa Notes
Animals, Chordates, Humans, Mammals, Primates, Vertebrates

L70 ANSWER 7 OF 25 BIOSIS COPYRIGHT (c) 2008 The Thomson Corporation on STN DUPLICATE 2

ACCESSION NUMBER: 2003:38315 BIOSIS Full-text
DOCUMENT NUMBER: PREV200300038315
TITLE: Biochemical screening for aneuploidy in ovum donor pregnancies.
AUTHOR(S): Donnenfeld, Alan E. [Reprint Author]; Icke, Katherine V.; Pargas, Carol; Dowman, Christine
CORPORATE SOURCE: Genzyme Genetics, Philadelphia, PA, USA
SOURCE: American Journal of Obstetrics and Gynecology, (November 2002) Vol. 187, No. 5, pp. 1222-1225. print.
CODEN: AJOGAH. ISSN: 0002-9378.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 8 Jan 2003
Last Updated on STN: 8 Jan 2003

ABSTRACT:OBJECTIVE: The purpose of this study was to compare the screening efficacy for aneuploidy detection in ovum donor pregnancies with the use of either the age of the ovum donor or the ovum recipient. STUDY DESIGN: Second-trimester biochemical screening for aneuploidy with alpha-fetoprotein, unconjugated estriol, and human chorionic gonadotropin

was performed on maternal serum samples that were submitted prospectively from singleton ovum donor pregnancies. The calculation of aneuploidy risks were performed separately with the age of the ovum donor or the ovum recipient. Risks of >1 in 295 and >1 in 100 were used as cutoff ***values*** for the identification of screen -positive pregnancies for Down syndrome and trisomy 18, respectively. RESULTS: Samples from 93 ovum donor pregnancies were identified. The mean ages of the ovum donors and recipients were 27 years (range 20-38.5 years) and 43.6 years (range, 25.9-54.3 years), respectively. When the age of the ovum donor was used in the determination of aneuploidy risk, there were 9 screen-positive pregnancies (9.7%), whereas the use of the age of the ovum recipient resulted in 76 screen-positive pregnancies (82%). With the use of the McNemar test for paired observations, the proportion of screen-positive pregnancies with the age of the ovum donor (9.7%) compared with the age of the ovum recipient (82%) was statistically significant (P<.0001). The odds of being affected, given a positive result, were 1 in 9 (11%) with the age of the ovum recipient and 1 in 76 (1.3%) with the age of the ovum donor. The only fetus with aneuploidy (trisomy 18) was identified as being screen positive in both the ovum donor and ovum recipient calculations. CONCLUSION: In ovum donor pregnancy aneuploidy risk calculations, the use of the age of the ovum donor instead of the ovum recipient reduces the false-positive rate and improves screening efficacy.

CONCEPT CODE: Genetics - Human 03508
Biochemistry studies - Proteins, peptides and amino acids 10064
Pathology - Diagnostic 12504
Reproductive system - Physiology and biochemistry 16504
Reproductive system - Pathology 16506
Endocrine - Gonads and placenta 17006
Development and Embryology - Pathology 25503

INDEX TERMS: Major Concepts
Medical Genetics (Allied Medical Sciences); Methods and Techniques; Obstetrics (Human Medicine, Medical Sciences)

INDEX TERMS: Parts, Structures, & Systems of Organisms
chromosome 18; ovum: reproductive system

INDEX TERMS: Diseases
aneuploidy: genetic disease, diagnosis
Aneuploidy (MeSH)

INDEX TERMS: Diseases
trisomy 18: congenital disease, genetic disease, diagnosis
Trisomy (MeSH)

INDEX TERMS: Chemicals & Biochemicals
alpha-fetoprotein; human chorionic gonadotropin [hCG]; unconjugated estriol

INDEX TERMS: Methods & Equipment
biochemical screening: clinical techniques

INDEX TERMS: Miscellaneous Descriptors
gestational age; pregnancy; risk assessment

ORGANISM: Classifier
Hominidae 86215
Super Taxa
Primates; Mammalia; Vertebrata; Chordata; Animalia
Organism Name
human (common): adult, ovum donor, ovum recipient, patient, female

Taxa Notes
Animals, Chordates, Humans, Mammals, Primates, Vertebrates
REGISTRY NUMBER: 9002-61-3 (human chorionic gonadotropin)
9002-61-3 (hCG)

L70 ANSWER 8 OF 25 EMBASE COPYRIGHT (c) 2008 Elsevier B.V. All rights reserved on STN DUPLICATE 3
ACCESSION NUMBER: 2003437722 EMBASE Full-text
TITLE: Combined ultrasound and biochemical screening for Down's Syndrome in the first trimester: A Scottish multicentre study.
AUTHOR: Crossley, Jennifer A.; Aitken, David A., Dr. (correspondence); McBride, Elizabeth; Connor, J. Michael
CORPORATE SOURCE: Institute of Medical Genetics, Yorkhill NHS Trust, Glasgow, United Kingdom.
AUTHOR: Cameron, Alan D.
CORPORATE SOURCE: Fetal Medicine Department, Yorkhill NHS Trust, Glasgow, United Kingdom.
AUTHOR: Aitken, David A., Dr. (correspondence)
CORPORATE SOURCE: Institute of Medical Genetics, Yorkhill, Glasgow G3 8SJ, United Kingdom.
SOURCE: BJOG: An International Journal of Obstetrics and Gynaecology, (Jun 2002) Vol. 109, No. 6, pp. 667-676.
Refs: 30
ISSN: 1470-0328 CODEN: BIOGFQ
PUBLISHER IDENT.: S 1470-0328(02)01394-0
COUNTRY: United Kingdom
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 010 Obstetrics and Gynecology
017 Public Health, Social Medicine and Epidemiology
029 Clinical and Experimental Biochemistry
008 Neurology and Neurosurgery
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 1 Dec 2003
Last Updated on STN: 1 Dec 2003
ABSTRACT: Objective: To evaluate the use of ultrasound measurements of fetal nuchal translucency (NT) obtained in a routine antenatal clinic setting in combination with appropriate biochemical markers as a first trimester screening test for Down's ***Syndrome.*** Design: Multicentre observational study. Setting: Fifteen Scottish maternity units. Population: Pregnant women (n = 17,229) attending routine antenatal clinics at 10-14 weeks of gestation. Methods: NT measurements were attempted in all women along with the measurement of maternal serum free beta human chorionic gonadotrophin (FβhCG) and pregnancy-associated plasma protein-A (PAPP-A). All results were converted to multiples of the appropriate gestational median (MoM) and using a statistical model the risk of an affected pregnancy was derived. No results were given to participating women but all were offered routine second trimester biochemical screening. All cases of Down's Syndrome within the study group were ascertained and the detection rate for each marker was estimated. Main outcome measures: Success rate of obtaining NT measurements and overall effectiveness of ultrasound and biochemical ***markers*** individually and in combination for the detection of Down's Syndrome pregnancies. Results:

NT measurements were obtained in 72.9% of women and blood samples in 98.4%. Forty-five cases of Down's Syndrome were ascertained (2.6/1000). NT measurements were obtained in 37 cases (median NT 1.65 MoM), blood samples in 42 cases and both NT and blood in 34 cases. In combination with the a priori maternal age risk, observed detection rates at a 5% false positive rate were 20/37 (54%) for NT, 23/42 (55%) for FβhCG and PAPP-A and 28/34 (82%) for a combination of NT, FβhCG and PAPP-A using a cutoff risk of 1:250. The effect of failing to obtain NT measurements in all cases reduces the overall detection rate to 62% (i.e. 28/45) if the entire series of affected pregnancies within the study group is considered. Conclusions: NT in combination with appropriate serum markers has the potential to detect over 80% of ***Down*** 's Syndrome fetuses in early ***pregnancy.*** However, NT measurement is highly operator-dependent. It requires training, external quality control and adequate time to allow accurate measurement, otherwise suboptimal performance will result.

CONTROLLED TERM: Medical Descriptors:
adult
article
blood sampling
chemical analysis
controlled study
diagnostic accuracy
diagnostic approach route
diagnostic value
*Down syndrome: DI, diagnosis
female
*fetus echography
*first trimester pregnancy
high risk pregnancy
hormone blood level
human
maternal age
maternal serum
prenatal diagnosis
*prenatal screening
priority journal
protein blood level
risk factor
second trimester pregnancy
statistical model
United Kingdom

CONTROLLED TERM: Drug Descriptors:
biochemical marker: EC, endogenous compound
chorionic gonadotropin beta subunit: EC, endogenous compound
pregnancy associated plasma protein A: EC, endogenous compound

L70 ANSWER 9 OF 25 MEDLINE on STN
ACCESSION NUMBER: 2001671014 MEDLINE Full-text
DOCUMENT NUMBER: PubMed ID: 11717625
TITLE: The impact of the use of the isolated echogenic intracardiac focus as a screen for Down syndrome in women under the age of 35 years.
AUTHOR: Caughey A B; Lyell D J; Filly R A; Washington A E; Norton M E
CORPORATE SOURCE: Department of Obstetrics, Gynecology & Reproductive

Sciences, University of California, San Francisco
94143, USA.. caugheya@obgyn.ucsf.edu
SOURCE: American journal of obstetrics and gynecology, (2001 Nov) Vol. 185, No. 5, pp. 1021-7.
Journal code: 0370476. ISSN: 0002-9378.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Abridged Index Medicus Journals; Priority Journals
ENTRY MONTH: 200112
ENTRY DATE: Entered STN: 22 Nov 2001
Last Updated on STN: 23 Jan 2002
Entered Medline: 19 Dec 2001

ABSTRACT:
OBJECTIVE: The purpose of this study was to determine the public health impact of the routine offering of amniocentesis to women under the age of 35 years who have an isolated fetal echogenic intracardiac focus on ***second*** trimester ultrasound scan. STUDY DESIGN: A decision analytic model was designed that compared the accepted standard of ***second*** trimester triple marker screen for ***Down*** syndrome to a policy in which amniocentesis with an isolated echogenic intracardiac focus on ultrasound in addition to the triple marker screen is offered to all women in the United States who are <35 years of age. A sensitivity of 20%, an echogenic intracardiac focus screen positive rate of 5%, and a risk of Down syndrome of 1:1000 were assumed. A sensitivity analysis was performed that varied the screen positive rate, the sensitivity of echogenic intracardiac focus for Down syndrome, and the prescreen risk for Down syndrome in the population. RESULTS: With the baseline sensitivities, rates, and risks, the use of isolated echogenic intracardiac focus as a screen would result in an additional 118,146 amniocenteses performed annually to diagnose 244 ***fetuses*** with Down syndrome. These amniocenteses would result in 582 additional miscarriages. It would be necessary to perform 485 amniocenteses that would result in 2.4 procedure-related losses for each additional Down ***syndrome*** fetus that was identified.
CONCLUSION: Although the echogenic intracardiac focus appears to be associated with a small increased risk of Down syndrome, its use as a screening tool in low-risk populations would lead to a large number of amniocenteses and miscarriages to identify a small number of Down ***syndrome*** fetuses.

CONTROLLED TERM: Check Tags: Female
Abortion, Spontaneous: EP, epidemiology
Abortion, Spontaneous: ET, etiology
Adult
Amniocentesis: AE, adverse effects
Amniocentesis: SN, statistics & numerical data
Decision Support Techniques
Down Syndrome: ET, etiology
*Down Syndrome: US, ultrasonography
*Fetal Heart: US, ultrasonography
Humans
Incidence
*Mass Screening: MT, methods
Pregnancy
Risk Factors
Sensitivity and Specificity
*Ultrasonography, Prenatal

L70 ANSWER 10 OF 25 MEDLINE on STN
ACCESSION NUMBER: 2001016148 MEDLINE Full-text
DOCUMENT NUMBER: PubMed ID: 10986181
TITLE: Participation in maternal serum screening for Down syndrome, neural tube defects, and trisomy 18 following screen-positive results in a previous pregnancy.
AUTHOR: Rausch D N; Lambert-Messerlian G M; Canick J A
CORPORATE SOURCE: Department of Pathology and Laboratory Medicine, Women and Infants Hospital, Brown University School of Medicine, 70 Elm St, Providence, RI 02903, USA.
SOURCE: The Western journal of medicine, (2000 Sep) Vol. 173, No. 3, pp. 180-3.
Journal code: 0410504. ISSN: 0093-0415.
COMMENT: Comment in: West J Med. 2000 Sep;173(3):183-4. PubMed ID: 10986182
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Abridged Index Medicus Journals; Priority Journals
ENTRY MONTH: 200010
ENTRY DATE: Entered STN: 22 Mar 2001
Last Updated on STN: 22 Mar 2001
Entered Medline: 27 Oct 2000

ABSTRACT:
OBJECTIVE: To determine whether women who have had a positive serum ***screening*** result for Down syndrome or neural tube defect in 1 pregnancy have a lower rate of participation in screening in their next pregnancy. SETTING: A triple-marker ***screening*** program at a university hospital. METHODS: Pregnancy and screening information was collected from laboratory and hospital databases to compare subsequent screening participation of women who were screen-negative and screen-positive for the risk of a fetus with Down syndrome or a neural tube defect. RESULTS: In an age-matched comparison, 108 women who had a previous screen-positive result were significantly less likely than 108 women who were screen-negative to participate in maternal serum screening in their next pregnancy. When examined according to the type of screen-positive result, the effect was significant for both those who were screen-positive for Down syndrome and those who were screen-positive for neural tube defect. The degree of risk in screen-positive women did not significantly affect their participation in screening in the next pregnancy. CONCLUSIONS: Anxiety related to a screen-positive result probably causes decreased participation in maternal serum screening in the next pregnancy. Reducing the screen-positive rate in prenatal serum screening would alleviate maternal anxiety and would probably lead to more stable participation.

CONTROLLED TERM: Check Tags: Female
Anxiety
*Biological Markers: BL, blood
Chi-Square Distribution
Chorionic Gonadotropin: BL, blood
*Chromosomes, Human, Pair 18
*Down Syndrome: DI, diagnosis
Estriol: BL, blood
Humans
Mass Screening: MT, methods
Mass Screening: PX, psychology
*Mass Screening: SN, statistics & numerical

data
*Neural Tube Defects: DI, diagnosis
*Patient Participation
Pregnancy
*Prenatal Diagnosis: MT, methods
Prenatal Diagnosis: PX, psychology
*Trisomy: DI, diagnosis
alpha-Fetoproteins: AN, analysis
CAS REGISTRY NO.: 50-27-1 (Estriol)
CHEMICAL NAME: 0 (Biological Markers); 0 (Chorionic Gonadotropin); 0 (alpha-Fetoproteins)

L70 ANSWER 11 OF 25 HCAPLUS COPYRIGHT 2008 ACS on STN
ACCESSION NUMBER: 2000:635650 HCAPLUS Full-text
DOCUMENT NUMBER: 134:191304
ENTRY DATE: Entered STN: 13 Sep 2000
TITLE: Biochemical screening for Down syndrome
AUTHOR(S): Cuckle, H.
CORPORATE SOURCE: 26 Clarendon Road, School of Medicine, Growth and Development, Centre for Reproduction, Reproductive Epidemiology, University of Leeds, Leeds, LS2 9NZ, UK
SOURCE: European Journal of Obstetrics & Gynecology and Reproductive Biology (2000), 92(1), 97-101
CODEN: EOGRAL; ISSN: 0301-2115
PUBLISHER: Elsevier Science Ireland Ltd.
DOCUMENT TYPE: Journal; General Review
LANGUAGE: English
CLASSIFICATION: 14-0 (Mammalian Pathological Biochemistry)
Section cross-reference(s): 2

ABSTRACT:
A review with 16 refs. Maternal serum screening for ***Down*** syndrome is an established practise in many countries. In the second trimester human chorionic gonadotropin (hCG) or free β-hCG is the marker of first choice, with α-fetoprotein (AFP) as the second marker and unconjugated estriol (uE3) the third. Statistical models with ***parameters*** derived by meta-anal. predict that a three ***marker*** combination will yield a 67% detection rate for a 5% false-pos. rate. The model prediction have been confirmed in 21 large prospective intervention studies. A fourth marker, inhibin A, increases the detection rate by 7% for the same false-pos. rate. In the first trimester, similar models predict that a combination of pregnancy associated plasma protein A, free β-hCG, AFP and uE3will yield a 70% detection rate. This is increased to 88% if ultrasound nuchal translucency is used as an addnl. marker . Screening can also be extended to Edwards' syndrome, yielding high detection rates with little increase in the false-pos. rate. Abnormal marker levels are also associated with a variety of adverse outcomes of pregnancy. High quality information and decision aids are needed to minimize anxiety among screenees.

SUPPL. TERM: review hormone biochem marker fetus diagnosis Down syndrome
INDEX TERM: Embryo, animal
(fetus; serum and urine biochem. markers for prenatal diagnosis of

Down syndrome in human)
INDEX TERM: Diagnosis
(prenatal; serum and urine biochem. markers for prenatal diagnosis of Down syndrome in human)
INDEX TERM: Biomarkers (biological responses)
Down's syndrome
Pregnancy
(serum and urine biochem. markers for prenatal diagnosis of Down syndrome in human)
INDEX TERM: α-Fetoproteins
ROLE: BAC (Biological activity or effector, except adverse); BSU (Biological study, unclassified); THU (Therapeutic use); BIOL (Biological study); USES (Uses)
(serum and urine biochem. markers for prenatal diagnosis of Down syndrome in human)
INDEX TERM: 50-27-1, Estriol 9002-61-3, Chorionic gonadotropin 102510-92-9, Inhibin A
ROLE: BAC (Biological activity or effector, except adverse); BSU (Biological study, unclassified); THU (Therapeutic use); BIOL (Biological study); USES (Uses)
(serum and urine biochem. markers for prenatal diagnosis of Down syndrome in human)
REFERENCE COUNT: 16 THERE ARE 16 CITED REFERENCES AVAILABLE FOR THIS RECORD.
REFERENCE(S):
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L70 ANSWER 12 OF 25 HCAPLUS COPYRIGHT 2008 ACS on STN
ACCESSION NUMBER: 1999:709007 HCAPLUS Full-text
DOCUMENT NUMBER: 131:319902
ENTRY DATE: Entered STN: 05 Nov 1999
TITLE: Antenatal screening for Down's syndrome
INVENTOR(S): Wald, Nicholas John
PATENT ASSIGNEE(S): UK
SOURCE: PCT Int. Appl., 45 pp.
CODEN: PIXXD2
DOCUMENT TYPE: Patent

LANGUAGE: English
INT. PATENT CLASSIF.:
MAIN: G01N033-68
SECONDARY: A61B008-08
CLASSIFICATION: 9-16 (Biochemical Methods)
Section cross-reference(s): 2, 14
FAMILY ACC. NUM. COUNT: 1
PATENT INFORMATION:

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
WO 9956132	A1	19991104	WO 1999-GB1341	19990429
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W: AE, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, CA, CH, CN, CU, CZ, DE, DK, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MD, MG, MK, MN, MW, MX, NO, NZ, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, UA, UG, US, UZ, VN, YU, ZA, ZW				
RW: GH, GM, KE, LS, MW, SD, SL, SZ, UG, ZW, AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG				
CA 2330538	A1	19991104	CA 1999-2330538	19990429
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CA 2330538	C	20070911		
AU 9936213	A	19991116	AU 1999-36213	19990429
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AU 763171	B2	20030717		
EP 1076824	A1	20010221	EP 1999-918188	19990429
			<--	
EP 1076824	B1	20060614		
R: AT, BE, CH, DE, DK, ES, FR, GB, GR, IT, LI, LU, NL, PT, FI				
US 6573103	B1	20030603	US 1999-301621	19990429
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IL 139302	A	20050725	IL 1999-139302	19990429
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AT 330226	T	20060715	AT 1999-918188	19990429
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PT 1076824	T	20061031	PT 1999-918188	19990429
			<--	
ES 2262319	T3	20061116	ES 1999-918188	19990429
			<--	

US 20030175981	A1	20030918	US 2003-389968	200303 18
			<--	
PRIORITY APPLN. INFO.:			GB 1998-9209	A 199804 29
			<--	
			GB 1998-13905	A 199806 26
			<--	
			US 1999-301621	A3 199904 29
			<--	
			WO 1999-GB1341	W 199904 29
			<--	

PATENT CLASSIFICATION CODES:

PATENT NO.	CLASS	PATENT FAMILY CLASSIFICATION CODES
WO 9956132	ICM	G01N033-68
	ICS	A61B008-08
	IPCI	G01N0033-68 [ICM,6]; A61B0008-08 [ICS,6]
	IPCR	G01N0033-68 [I,C*]; G01N0033-68 [I,A]
	ECLA	G01N033/68T; S01N
CA 2330538	IPCI	A61B0008-08 [N,A]; G01N0033-68 [I,A]; G01N0033-74 [I,A]; G01N0033-76 [I,A]
	IPCR	G01N0033-74 [I,C]; G01N0033-76 [I,A]; A61B0008-08 [N,C]; A61B0008-08 [N,A]; G01N0033-68 [I,C]; G01N0033-68 [I,A]; G01N0033-74 [I,A]
	ECLA	G01N033/68T; S01N
AU 9936213	IPCI	G01N0033-68 [ICM,6]; A61B0008-08 [ICS,6]
	IPCR	G01N0033-68 [I,C*]; G01N0033-68 [I,A]
	ECLA	G01N033/68T; S01N
EP 1076824	IPCI	A61B0008-08 [I,C]; G01N0033-68 [I,C]; G01N0033-68 [I,A]; A61B0008-08 [I,A]
	IPCR	G01N0033-68 [I,C*]; G01N0033-68 [I,A]
	ECLA	G01N033/68T; S01N
US 6573103	IPCI	G01N0033-48 [ICM,7]
	IPCR	G01N0033-68 [I,C*]; G01N0033-68 [I,A]
	NCL	436/065.000; 435/004.000; 436/086.000; 436/510.000; 436/814.000; 436/818.000
	ECLA	G01N033/68T; S01N
IL 139302	IPCI	G01N0033-68 [ICM,7]
	IPCR	G01N0033-68 [I,C*]; G01N0033-68 [I,A]
	ECLA	G01N033/68T
AT 330226	IPCI	G01N0033-68 [ICS,7]; A61B0008-08 [ICS,7]
	IPCR	G01N0033-68 [I,C*]; G01N0033-68 [I,A]
	ECLA	G01N033/68T
PT 1076824	IPCI	G01N0033-68 [ICS,7]
	IPCR	G01N0033-68 [I,C*]; G01N0033-68 [I,A]
	ECLA	G01N033/68T
ES 2262319	IPCI	G01N0033-68 [I,C]; A61B0008-08 [I,C]; G01N0033-68 [I,A]; A61B0008-08 [I,A]
	IPCR	G01N0033-68 [I,C]; G01N0033-68 [I,A]; A61B0008-08 [I,C]; A61B0008-08 [I,A]
	ECLA	G01N033/68T

US 20030175981 IPCI G01N0033-53 [ICM,7]
IPCR G01N0033-68 [I,C*]; G01N0033-68 [I,A]
NCL 436/065.000; 436/086.000; 436/510.000
ECLA G01N033/68T; S01N

ABSTRACT:
A method of screening for fetal Down's ***syndrome*** is described. Screening marker levels are measured. These may be measurements of a biochem. ***marker*** in a maternal sample or measurements of a marker from an ultrasound scan. The marker levels are used to calculate a risk of Down's syndrome. Instead of using markers from a single stage of pregnancy, the method uses markers from two or more different stages of pregnancy, typically one being in the first and another being in second trimester. The method may be automated.

SUPPL. TERM: antenatal screening Down syndrome
INDEX TERM: Diagnosis
(Antenatal; antenatal screening for Down's syndrome)
INDEX TERM: Parturition
(Multiple; antenatal screening for Down's syndrome)
INDEX TERM: Blood analysis
Body weight
Diabetes mellitus
Down's syndrome
Multivariate analysis
Pregnancy
Refrigeration
Sound and Ultrasound
Urine analysis
(antenatal screening for Down's syndrome)
INDEX TERM: α-Fetoproteins
ROLE: ANT (Analyte); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(antenatal screening for Down's syndrome)
INDEX TERM: Embryo, animal
(fetus; antenatal screening for Down's syndrome)
INDEX TERM: Statistical analysis
(multivariate Gaussian anal.; antenatal screening for Down's syndrome)
INDEX TERM: 9002-61-3, Human chorionic gonadotropin 9002-61-3D, Human chorionic gonadotropin, beta-subunit derivs. 56832-30-5 102510-92-9, Inhibin a 151662-33-8
ROLE: ANT (Analyte); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(antenatal screening for Down's syndrome)
INDEX TERM: 50-27-1, Estriol
ROLE: ANT (Analyte); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(unconjugated; antenatal screening for

Down's syndrome)
REFERENCE COUNT: 2 THERE ARE 2 CITED REFERENCES AVAILABLE FOR THIS RECORD.
REFERENCE(S): (1) Ciba Corning Diagnostics Corp; WO 9703363 A 1997 HCAPLUS
(2) Wald, N; Annals of Medicine 1994, V26(1), P23 MEDLINE

L70 ANSWER 13 OF 25 HCAPLUS COPYRIGHT 2008 ACS on STN
ACCESSION NUMBER: 2000:29672 HCAPLUS Full-text
DOCUMENT NUMBER: 132:331614
ENTRY DATE: Entered STN: 13 Jan 2000
TITLE: Maternal serum superoxide dismutase (SOD): a possible marker for screening Down syndrome affected pregnancies
AUTHOR(S): Ognibene, Agostino; Ciuti, Riccardo; Tozzi, Paola; Messeri, Gianni
CORPORATE SOURCE: Laboratory of Clinical Biochemistry, Azienda Ospedaliera Careggi, Florence, 50139, Italy
SOURCE: Prenatal Diagnosis (1999), 19(11), 1058-1060
CODEN: PRDIDM; ISSN: 0197-3851
PUBLISHER: John Wiley & Sons Ltd.
DOCUMENT TYPE: Journal
LANGUAGE: English
CLASSIFICATION: 9-16 (Biochemical Methods)
Section cross-reference(s): 7, 13, 14
ABSTRACT:
Superoxide dismutase (SOD: EC 1.15.1.1) has been shown to increase in Down syndrome (DS) subjects and in amniotic fluid from DS affected pregnancies. In order to verify a possible increase of maternal serum SOD in DS affected pregnancies and its possible contribution in prenatal ***screening*** , the serum enzyme activity was retrospectively measured in samples from normal and DS affected pregnancies. Alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), unconjugated oestriol (uE3) and serum SOD were measured in serum samples collected from 80 normal and 9 DS affected second-trimester pregnancies. The maternal serum SOD activity in the DS group (3.12 ± 0.73 U/mL) was significantly higher ($p < 0.001$) than in the control one (2.20 ± 0.7 U/mL). The addition of SOD appeared to be capable of improving the sensitivity of the conventional multi-parametric test (AFP, uE3 and hCG) even if the small number of subjects did not allow the achievement of statistical significance.

SUPPL. TERM: superoxide dismutase blood mother diagnosis
Down syndrome human
INDEX TERM: Embryo, animal
(fetus; maternal serum superoxide dismutase (SOD) as possible marker for screening Down syndrome affected pregnancies)
INDEX TERM: Blood analysis
Blood serum
Diagnosis
Down's syndrome
Pregnancy
(maternal serum superoxide dismutase (SOD) as possible marker for screening Down syndrome affected

pregnancies)
INDEX TERM: α-Fetoproteins
ROLE: ANT (Analyte); BOC (Biological occurrence); BSU (Biological study, unclassified); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); OCCU (Occurrence); USES (Uses)
(maternal serum superoxide dismutase (SOD) as possible marker for screening Down syndrome affected pregnancies)
INDEX TERM: Diagnosis
(prenatal; maternal serum superoxide dismutase (SOD) as possible marker for screening Down syndrome affected pregnancies)
INDEX TERM: 50-27-1, Estriol 9002-61-3, Human chorionic gonadotropin 9054-89-1, Superoxide dismutase
ROLE: ANT (Analyte); BOC (Biological occurrence); BSU (Biological study, unclassified); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); OCCU (Occurrence); USES (Uses)
(maternal serum superoxide dismutase (SOD) as possible marker for screening Down syndrome affected pregnancies)
REFERENCE COUNT: 13 THERE ARE 13 CITED REFERENCES AVAILABLE FOR THIS RECORD.
REFERENCE(S): (1) Baeteman, M; Acta Paediatr Scand 1985, V74, P697 MEDLINE
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(11) Sinet, P; Exp Cell Res 1976, V97, P47 HCAPLUS
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(13) Tan, Y; J Exp Med 1973, V137, P317 HCAPLUS

L70 ANSWER 14 OF 25 EMBASE
ACCESSION NUMBER: 1999353556 EMBASE Full-text
TITLE: The proform of eosinophil major basic protein: A new maternal serum marker for Down syndrome.
AUTHOR: Christiansen, Michael (correspondence); Qin, Qiu-Ping; Nguyen, Tri H.; Norgaard-Pedersen, Bent
CORPORATE SOURCE: Department of Clinical Biochemistry, Statens Serum Institut, 5 Artillerivej, Copenhagen DK 2300 S, Denmark. mic@ssi.dk
AUTHOR: Oxvig, Claus; Overgaard, Michael T.; Sottrup-Jensen, Lars
CORPORATE SOURCE: Dept. of Molec. and Struct. Biology, University of Aarhus, Aarhus, Denmark.
AUTHOR: Wagner, Jill M.; Gleich, Gerald J.

CORPORATE SOURCE: Depts. of Immunology and Medicine, Mayo Clinic and Foundation, Rochester, MN, United States.
AUTHOR: Larsen, Severin O.
CORPORATE SOURCE: Department of Biostatistics, Statens Serum Institut, Copenhagen, Denmark.
SOURCE: Prenatal Diagnosis, (1999) Vol. 19, No. 10, pp. 905-910.
Refs: 48
ISSN: 0197-3851 CODEN: PRDIDM
COUNTRY: United Kingdom
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 010 Obstetrics and Gynecology
022 Human Genetics
008 Neurology and Neurosurgery
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 29 Oct 1999
Last Updated on STN: 29 Oct 1999

ABSTRACT: The proform of eosinophil major basic protein (proMBP), the most abundant protein in the eosinophil specific granule, is synthesized by the placenta and secreted into the maternal circulation, where it is found complex-bound to pregnancy-associated plasma protein-A (PAPP-A) and other proteins. We examined the potential of proMBP as a maternal serum ***marker*** for fetal Down syndrome (DS) by ***determining*** its maternal serum concentration (MSpMBP) in 25 ***Down*** syndrome (DS) pregnancies and 152 control pregnancies in the first trimester, and in 105 DS pregnancies and 156 control pregnancies in the second trimester. The median (95 per cent confidence interval) MSpMBP MoM in DS pregnancies (n = 15) was 0.66 (0.49-0.79) in gestational weeks 5-9; 1.06 (0.71-1.97) in weeks 10-12 (n = 10) and 1.62 (1.18-1.98) in weeks 14-20 (n = 105). Using parameterized receiver operator characteristics analysis for proMBP as a single marker for DS, ***detection*** rates (DRs) of 22 per cent and 38 per cent, for false-positive rates (FPRs) of 5 per cent, were found in weeks 5-9 (using MSpMBP ≤ cut-off) and weeks 14-20 (using MSpMBP ≥ cut-off), respectively. When age and MSpMBP were used as markers in combination, a DR of 36.8 per cent for an FPR of 5.5 per cent was obtained in weeks 5-9 using a risk cut-off of 1:250. In weeks 14-20 the DR was 48.4 per cent for an FPR of 5.3 per cent using the same risk cut-off. This makes proMBP a marker comparable in ***diagnostic*** efficiency to human chorionic gonadotrophin (hCG), and exceeding that of alpha-fetoprotein (AFP) and unconjugated oestriol (uE3), in the second trimester.

CONTROLLED TERM: Medical Descriptors:
adult
article
blood level
comparative study
controlled study
diagnostic accuracy
*Down syndrome: CN, congenital disorder
*Down syndrome: DI, diagnosis
female
fetus malformation: CN, congenital disorder
fetus malformation: DI, diagnosis
first trimester pregnancy
gestational age
human

human cell
major clinical study
*maternal serum
prenatal screening
priority journal
second trimester pregnancy
statistical analysis

CONTROLLED TERM: Drug Descriptors:
alpha fetoprotein: EC, endogenous compound
biological marker: EC, endogenous compound
chorionic gonadotropin: EC, endogenous compound
estriol: EC, endogenous compound
*major basic protein: EC, endogenous compound

CAS REGISTRY NO.: (chorionic gonadotropin) 9002-61-3; (estriol) 50-27-1

L70 ANSWER 15 OF 25 EMBASE COPYRIGHT (c) 2008 Elsevier B.V. All rights reserved on STN

ACCESSION NUMBER: 1999205922 EMBASE Full-text
TITLE: Maternal serum screening for Down syndrome in the first trimester: Experience from Belarus.
AUTHOR: Tsukerman, G.L. (correspondence); Gusina, N.B.
CORPORATE SOURCE: Institute for Hereditary Diseases, Centre for Medical Genetic Services, Minsk, Belarus.
AUTHOR: Tsukerman, G.L. (correspondence)
CORPORATE SOURCE: Institute for Hereditary Diseases, Centre for Medical Genetic Services, Building 9, 66 Orlovskaya Street, Minsk 220053, Belarus.
AUTHOR: Cuckle, H.S.
SOURCE: Prenatal Diagnosis, (1999) Vol. 19, No. 6, pp. 499-504.
Refs: 20
ISSN: 0197-3851 CODEN: PRDIDM
COUNTRY: United Kingdom
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 010 Obstetrics and Gynecology
027 Biophysics, Bioengineering and Medical Instrumentation
008 Neurology and Neurosurgery
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 1 Jul 1999
Last Updated on STN: 1 Jul 1999

ABSTRACT: We have carried out a large retrospective study of α-fetoprotein (AFP), free-β human chorionic gonadotrophin (hCG) and pregnancy-associated plasma protein (PAPP-A) in the ***first*** trimester of pregnancy. Unlike other studies all women had routine ultrasound dating, carried out during a nuchal translucency measurement project. A total of 13,477 serum samples were tested for AFP and 11,659 for free β-hCG, A subset of 1564 samples from unaffected pregnancies were also tested for PAPP-A on a case-control basis. All three markers were also determined in 31 samples from ***pregnancies*** with Down syndrome. Equations were derived to express results in multiples of the median using both gestational age and crown-rump length and to adjust for maternal weight. ***Statistical*** modelling with Gaussian distribution ***parameters*** obtained in the study were used to predict the detection rate for a 5 per cent false-positive rate. The predicted rates were: 73.7 per cent for all three markers; 69.1 per cent for

PAPP-A and free β-hCG; 47.4 per cent for PAPP-A and AFP; 57.6 per cent for free β-hCG and AFP. As these rates are similar to those in the second trimester, health planners may now want to consider a change in policy from second-trimester to first -trimester screening with biochemical markers.

CONTROLLED TERM: Medical Descriptors:
adult
article
Belarus
controlled study
crown rump length
diagnostic error
*Down syndrome: CN, congenital disorder
*Down syndrome: DI, diagnosis
enzyme immunoassay
fetus
fetus echography
*first trimester pregnancy
fluorescent antibody technique
gestational age
hormone blood level
human
human cell
mathematical analysis
*prenatal screening
priority journal
retrospective study
second trimester pregnancy
statistical model
ultrasound
CONTROLLED TERM: Drug Descriptors:
alpha fetoprotein: EC, endogenous compound
biological marker: EC, endogenous compound
chorionic gonadotropin beta subunit: EC, endogenous compound
pregnancy associated plasma protein a: EC, endogenous compound
NAME OF PRODUCT: (1) DELFIA; (2) DIAplus-Roche
COMPANY NAME: (1) eg and g wallac oy (Finland) ; (2) Hoffmann La Roche (Russian Federation)

L70 ANSWER 16 OF 25 HCAPLUS COPYRIGHT 2008 ACS on STN
ACCESSION NUMBER: 1998:299210 HCAPLUS Full-text
DOCUMENT NUMBER: 129:80190
ORIGINAL REFERENCE NO.: 129:16549a,16552a
ENTRY DATE: Entered STN: 22 May 1998
TITLE: Second trimester maternal dimeric inhibin-A in the multiple-marker screening test for Down's syndrome
AUTHOR(S): Renier, Martin A.; Vereecken, Annie; Van Herck, Erik; Straetmans, Danny; Ramaekers, Paul; Buytaert, Philippe
CORPORATE SOURCE: University Hospital of Antwerp, Department of Obstetrics and Gynecology, University of Antwerp, Antwerp, Belg.
SOURCE: Human Reproduction (1998), 13(3), 744-748
CODEN: HUREEE; ISSN: 0268-1161
PUBLISHER: Oxford University Press

DOCUMENT TYPE: Journal
LANGUAGE: English
CLASSIFICATION: 14-14 (Mammalian Pathological Biochemistry)
ABSTRACT:
The aim of this study was to evaluate the addnl. value of dimeric inhibin-A serum concentration in second trimester multiple-***marker*** screening tests for pregnancies affected by Down's syndrome. The authors anticipated that second trimester maternal serum dimeric inhibin-A concns. would be altered in pregnancies complicated by fetal Down's ***syndrome*** and that dimeric inhibin-A would perform better than one of the three substances analyzed in the multiple-marker ***screening*** test currently in use. A total of 1156 serum samples were screened for dimeric inhibin-A in parallel with the routine classic triple test screening program performed on a random obstetric population. Classic triple test performance was compared with detection rates obtained after substitution of unconjugated estriol by inhibin-A and with the performance of inhibin-A and α-fetoprotein alone. Absolute dimeric inhibin-A maternal serum concns. of Down's syndrome ***pregnancies*** were indeed higher than those of normal pregnancies in the authors' screened population. The performance of dimeric inhibin-A in combination with the multiple-marker ***screening*** test, however, is limited because of its strong correlation with intact human chorionic gonadotropin.

SUPPL. TERM: blood inhibin A Down syndrome
fetus
INDEX TERM: Embryo, animal
(fetus; second trimester maternal dimeric inhibin-A in multiple-marker screening test for Down's syndrome in humans)
INDEX TERM: Blood serum
(maternal; second trimester maternal dimeric inhibin-A in multiple-marker screening test for Down's syndrome in humans)
INDEX TERM: Diagnosis
(prenatal; second trimester maternal dimeric inhibin-A in multiple-marker screening test for Down's syndrome in humans)
INDEX TERM: Down's syndrome
Pregnancy
(second trimester maternal dimeric inhibin-A in multiple-marker screening test for Down's syndrome in humans)
INDEX TERM: 102510-92-9, Inhibin A
ROLE: BOC (Biological occurrence); BSU (Biological study, unclassified); THU (Therapeutic use); BIOL (Biological study); OCCU (Occurrence); USES (Uses)
(second trimester maternal dimeric inhibin-A in multiple-marker screening test for Down's syndrome in humans)
REFERENCE COUNT: 27 THERE ARE 27 CITED REFERENCES AVAILABLE FOR THIS RECORD.
REFERENCE(S): (1) Aitken, D; N Engl J Med 1996, V334, P1231 MEDLINE
(2) Brambati, B; Br J Obstet Gynaecol 1993, V100, P324 MEDLINE

(3) Brock, D; Prenat Diagn 1990, V10, P245 MEDLINE
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(24) Wald, N; Br J Obstet Gynaecol 1991, V98, P905 MEDLINE
(25) Wald, N; Prenat Diagn 1997, V17, P285 MEDLINE
(26) Wallace, E; Clin Endocrinol 1996, V44, P17 MEDLINE
(27) Wallace, E; Prenat Diagn 1995, V15, P359 MEDLINE

L70 ANSWER 17 OF 25 MEDLINE on STN
ACCESSION NUMBER: 1998265186 MEDLINE Full-text
DOCUMENT NUMBER: PubMed ID: 9602476
TITLE: Preliminary evidence for associations between second-trimester human chorionic gonadotropin and unconjugated oestriol levels with pregnancy outcome in Down syndrome pregnancies.
AUTHOR: Benn P A
CORPORATE SOURCE: Department of Pediatrics, University of Connecticut Health Center, Farmington, CT 06030-6140, USA.
SOURCE: Prenatal diagnosis, (1998 Apr) Vol. 18, No. 4, pp. 319-24.
Journal code: 8106540. ISSN: 0197-3851.
PUB. COUNTRY: ENGLAND: United Kingdom
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 199807
ENTRY DATE: Entered STN: 31 Jul 1998
Last Updated on STN: 31 Jul 1998
Entered Medline: 17 Jul 1998

ABSTRACT:
Fifty-six cases of Down syndrome were ***identified*** in a population of women who had undergone maternal serum triple marker screening [alpha-fetoprotein

(AFP), human chorionic gonadotropin (hCG), and unconjugated oestriol (uE3) analyses]. These affected pregnancies represented all known cases present in the population of 34,368 women screened. Using a 1:270 mid-trimester Down syndrome risk to define the screen-positive group, 42 affected pregnancies were screen-positive (medians: AFP = 0.79 MOM, hCG = 2.13 MOM, uE3 = 0.62 MOM, age 34.6 years) and 14 pregnancies were screen-negative (medians: AFP = 0.82 MOM, hCG = 1.57 MOM, uE3 = 0.92 MOM, age 24.2 years). Four affected pregnancies were associated with in utero death and each of these cases was associated with relatively extreme ***values*** of AFP, hCG, and uE3, including the three highest levels of hCG in the entire series of Down syndrome ***pregnancies.*** Twenty-nine (15 screen-positive and 14 screen-negative) affected pregnancies resulted in liveborns. ***Down*** syndrome pregnancies had a significantly shorter gestational term than controls, and Down syndrome babies were also lighter than controls, even after adjustment for sex and gestational age. In affected pregnancies, a low uE3 level appeared to be associated with a greater chance of a small-for-gestational age baby. No correlations could be demonstrated between AFP or hCG levels and gestational age-adjusted term weight. Based on this small series, it would appear that uE3 may be particularly useful in detecting those Down syndrome cases associated with small-for-gestational age fetuses. A very high hCG ***value*** may indicate a higher probability of fetal death.

CONTROLLED TERM: Check Tags: Female
*Chorionic Gonadotropin: BL, blood
*Down Syndrome: BL, blood
Down Syndrome: DI, diagnosis
*Estriol: BL, blood
Fetal Death
Gestational Age
Humans
Pregnancy
*Pregnancy Outcome
Pregnancy Trimester, Second
Prenatal Diagnosis
alpha-Fetoproteins: AN, analysis

CAS REGISTRY NO.: 50-27-1 (Estriol)

CHEMICAL NAME: 0 (Chorionic Gonadotropin); 0 (alpha-Fetoproteins)

L70 ANSWER 18 OF 25 MEDLINE on STN

ACCESSION NUMBER: 1998245836 MEDLINE Full-text

DOCUMENT NUMBER: PubMed ID: 10178803

TITLE: Down syndrome serum marker screening: decision criteria and implicit values.

AUTHOR: Seror V; Costet N

CORPORATE SOURCE: Center of Health Economics Research, INSERM Unit 357-CNRS ERS 387, Hopital de Bicetre, Cedex, France.. seror@kb.inserm.fr

SOURCE: Health policy (Amsterdam, Netherlands), (1998 Jan) Vol. 43, No. 1, pp. 83-96.
Journal code: 8409431. ISSN: 0168-8510.

PUB. COUNTRY: Ireland

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE) (RESEARCH SUPPORT, NON-U.S. GOV'T)

LANGUAGE: English

FILE SEGMENT: Health

ENTRY MONTH: 199806

ENTRY DATE: Entered STN: 23 Feb 2001

Last Updated on STN: 23 Feb 2001
Entered Medline: 10 Jun 1998

ABSTRACT:
Maternal serum markers assess the individual risk of giving birth to a fetus with Down syndrome. Because this information is a probability, and because of the infinite number of cut-off risks that can be adopted, a decision criterion is needed to define a population screening program. While a decision criterion for cut-off risks may refer to arbitrations between risks, another criterion must be considered. This criterion focuses on a societal perspective by comparing the costs of the program to the expected benefits. We will first discuss the questions that are raised when assessing, in terms of cost-effectiveness, the consequences of having adopted the policy maker's preferences for prenatal diagnosis referral. Subsequently, we will discuss the implicit ***values*** attributed to the outcomes of the program when the societal point of view is reduced to societal profitability. This is accomplished by means of a cost-benefit analysis using the 'avoided costs' approach. The consequences of screening with 'double' and 'triple' tests were assessed using a database made of 10,108 observations, including 63 Down syndrome cases. For a cut-off risk of 1:250 (resulting in a 7% amniocentesis referral rate, regardless of the technique), conclusions in terms of decision making differ according to the effectiveness indicator. Although a criterion based on resource allocation would promote the triple test, cost-benefit analysis points out the impact on results of factors such as the amniocentesis related fetal losses or the introduction of equity principles.

CONTROLLED TERM: Check Tags: Female
Adult
Amniocentesis: AE, adverse effects
Amniocentesis: EC, economics
Amniocentesis: UT, utilization
*Biological Markers
Cost-Benefit Analysis
Decision Making
*Diagnostic Tests, Routine: EC, economics
Diagnostic Tests, Routine: ST, standards
*Down Syndrome: DI, diagnosis
France
*Health Care Rationing: EC, economics
Health Policy
Humans
Maternal-Fetal Exchange
Outcome Assessment (Health Care)
Pregnancy
*Prenatal Diagnosis: EC, economics
Prenatal Diagnosis: ST, standards
Risk Assessment
Social Values

CHEMICAL NAME: 0 (Biological Markers)

L70 ANSWER 19 OF 25 BIOSIS COPYRIGHT (c) 2008 The Thomson Corporation on STN
ACCESSION NUMBER: 1997:138578 BIOSIS Full-text
DOCUMENT NUMBER: PREV199799437781
TITLE: Do morphometric markers increase identification of Down's syndrome fetuses in an otherwise normal sonogram?.
AUTHOR(S): Lanouette, J. M. [Reprint author]; Quintero, R. A.;

Treadwell, M. C.; Johnson, M. P.; Carreno, C. A.; Kruger, M.; Wolfe, H. M.

CORPORATE SOURCE: Dep. Obstetrics Gynecol., Div. Maternal-Fetal Med., Hutzel Hosp., Detroit, MI, USA

SOURCE: American Journal of Obstetrics and Gynecology, (1997) Vol. 176, No. 1 PART 2, pp. S69.
Meeting Info.: 17th Annual Clinical, Scientific, and Business Meeting of the Society of Perinatal Obstetricians. Anaheim, California, USA. January 20-25, 1997.
CODEN: AJOGAH. ISSN: 0002-9378.

DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
Conference; (Meeting Poster)

LANGUAGE: English

ENTRY DATE: Entered STN: 2 Apr 1997
Last Updated on STN: 2 Apr 1997

CONCEPT CODE: General biology - Symposia, transactions and proceedings 00520
Genetics - Human 03508
Mathematical biology and statistical methods 04500
Radiation biology - Radiation and isotope techniques 06504
Behavioral biology - Human behavior 07004
Anatomy and Histology - Radiologic anatomy 11106
Pathology - Diagnostic 12504
Reproductive system - General and methods 16501
Reproductive system - Anatomy 16502
Reproductive system - Physiology and biochemistry 16504
Reproductive system - Pathology 16506
Bones, joints, fasciae, connective and adipose tissue - General and methods 18001
Bones, joints, fasciae, connective and adipose tissue - Anatomy 18002
Bones, joints, fasciae, connective and adipose tissue - Physiology and biochemistry 18004
Bones, joints, fasciae, connective and adipose tissue - Pathology 18006
Nervous system - General and methods 20501
Nervous system - Anatomy 20502
Nervous system - Physiology and biochemistry 20504
Nervous system - Pathology 20506
Psychiatry - Mental retardation 21006
Development and Embryology - Descriptive teratology and teratogenesis 25552
Public health - Public health administration and statistics 37010
Public health - Health services and medical care 37012

INDEX TERMS: Major Concepts
Behavior; Development; Genetics; Mathematical Biology (Computational Biology); Morphology; Nervous System (Neural Coordination); Neurology (Human Medicine, Medical Sciences); Pathology; Psychiatry (Human Medicine, Medical Sciences); Public Health (Allied Medical Sciences); Radiology (Medical Sciences); Reproductive System (Reproduction); Skeletal System (Movement and Support)

INDEX TERMS: Miscellaneous Descriptors
ADULT; BIPARIETAL DIAMETER; CONGENITAL DISEASE; DIAGNOSTIC METHOD; DOWN'S SYNDROME; FEMALE; FEMUR LENGTH; FETUS; KARYOTYPE; MORPHOMETRIC MARKER; NERVOUS SYSTEM DISEASE; NEUROLOGY; OBSTETRICS; PATIENT; PRENATAL DIAGNOSIS; RADIOLOGY; SONOGRAM; STATISTICAL ANALYSIS; TRANSCEREBELLAR DIAMETER; ULTRASOUND

ORGANISM: Classifier
Hominidae 86215
Super Taxa
Primates; Mammalia; Vertebrata; Chordata; Animalia
Organism Name
human
Taxa Notes
Animals, Chordates, Humans, Mammals, Primates, Vertebrates

L70 ANSWER 20 OF 25 BIOSIS COPYRIGHT (c) 2008 The Thomson Corporation on STN

ACCESSION NUMBER: 1996:73636 BIOSIS Full-text
DOCUMENT NUMBER: PREV199698645771
TITLE: Fetal heart rate patterns in pregnancies with chromosomal disorders or subsequent fetal loss.
AUTHOR(S): Martinez, Josep M. [Reprint author]; Comas, Carme; Ojuel, Julia; Borrell, Antoni; Puerto, Bienvenido; Fortuny, Albert
CORPORATE SOURCE: C/Galileo 134 2!o 2!a, Barcelona 08028, Spain
SOURCE: Obstetrics and Gynecology, (1996) Vol. 87, No. 1, pp. 118-121.
CODEN: OBGNAS. ISSN: 0029-7844.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 27 Feb 1996
Last Updated on STN: 27 Feb 1996

ABSTRACT:Objective: To evaluate whether an abnormal fetal heart rate (FHR) is associated with chromosomal abnormalities in ***pregnant*** women undergoing an invasive procedure for prenatal diagnosis, and to investigate an abnormal FHR's potential value in predicting fetal loss in chromosomally normal pregnancies after the procedure. Methods: This was a prospective study including 867 women, all consecutive singleton pregnancies at 10-18 weeks' gestation, who underwent chorionic villus sampling (n = 371) or genetic amniocentesis (n = 496) at our institution. Fetal heart rate, expressed as beats per minute, was measured before the invasive procedure. Structural malformations detected by ultrasound were excluded. Results: ***Chromosomal*** abnormalities were diagnosed in 25 fetuses, including 11 with trisomy 21. In ten of 25 ***fetuses*** with chromosomal abnormalities, the FHR was between the fifth and 95th percentiles established before the procedure for the chromosomally normal group with normal outcome. Using the fifth percentile as a cutoff for trisomy 21, the detection rate was 63.6%, with a specificity of 96.2% and a positive predictive ***value*** of 17.9% (one in 5.5) in our population. Moreover, in six of the ten chromosomally normal miscarriages occurring within 4 weeks after the procedure, the FHR was above the 95th percentile. Conclusion: Although the value of a single measurement for ***screening*** purposes needs to be confirmed by further investigation, our preliminary data suggest that chromosomal anomalies, especially trisomy 21, may be suspected in fetuses with an

abnormally low FHR in early pregnancy. In chromosomally normal ***fetuses*** , the detection of an abnormally high FHR in some degree may be predictive of fetal loss after the invasive procedure.

CONCEPT CODE: Cytology - Human 02508
Genetics - Human 03508
Mathematical biology and statistical methods 04500
Behavioral biology - Human behavior 07004
Biophysics - Methods and techniques 10504
Pathology - Diagnostic 12504
Cardiovascular system - Physiology and biochemistry 14504
Cardiovascular system - Heart pathology 14506
Blood - Other body fluids 15010
Reproductive system - Physiology and biochemistry 16504
Reproductive system - Pathology 16506
Nervous system - Physiology and biochemistry 20504
Nervous system - Pathology 20506
Psychiatry - Mental retardation 21006
Development and Embryology - Pathology 25503
Development and Embryology - Descriptive teratology and teratogenesis 25552
Public health - Public health administration and statistics 37010
Public health - Health services and medical care 37012

INDEX TERMS: Major Concepts
Behavior; Cardiovascular Medicine (Human Medicine, Medical Sciences); Cardiovascular System (Transport and Circulation); Cell Biology; Development; Genetics; Mathematical Biology (Computational Biology); Methods and Techniques; Nervous System (Neural Coordination); Neurology (Human Medicine, Medical Sciences); Pathology; Physiology; Psychiatry (Human Medicine, Medical Sciences); Public Health (Allied Medical Sciences); Reproductive System (Reproduction)

INDEX TERMS: Miscellaneous Descriptors
CHORIONIC VILLUS SAMPLING; CHROMOSOMAL ANOMALY; CHROMOSOMALLY NORMAL MISCARRIAGE; FETAL HEART RATE PATTERN ABNORMALITY; FETUS; GENETIC AMNIOCENTESIS; INVASIVE PROCEDURE; PRENATAL DIAGNOSIS; STATISTICAL ANALYSIS; TRISOMY 21

ORGANISM: Classifier
Hominidae 86215
Super Taxa
Primates; Mammalia; Vertebrata; Chordata; Animalia
Organism Name
human
Taxa Notes
Animals, Chordates, Humans, Mammals, Primates, Vertebrates

L70 ANSWER 21 OF 25 MEDLINE on STN
ACCESSION NUMBER: 1995153455 MEDLINE Full-text
DOCUMENT NUMBER: PubMed ID: 7850586
TITLE: Does gender have an impact on the sonographic detection of second-trimester fetuses with Down's

syndrome?.
AUTHOR: Benacerraf B R; Miller W A; Nadel A; Pauker S; Bromley B
CORPORATE SOURCE: Department of Obstetrics and Gynecology, Brigham and Women's Hospital and Massachusetts General Hospital, Boston.
SOURCE: Ultrasound in obstetrics & gynecology : the official journal of the International Society of Ultrasound in Obstetrics and Gynecology, (1995 Jan) Vol. 5, No. 1, pp. 30-3.
Journal code: 9108340. ISSN: 0960-7692.
PUB. COUNTRY: ENGLAND: United Kingdom
DOCUMENT TYPE: (CLINICAL TRIAL)
(COMPARATIVE STUDY)
Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 199503
ENTRY DATE: Entered STN: 22 Mar 1995
Last Updated on STN: 22 Mar 1995
Entered Medline: 13 Mar 1995

ABSTRACT:
The biometric and structural sonographic features of 95 second -trimester fetuses with Down's syndrome were evaluated to determine whether affected male ***fetuses*** differed from affected females. There were 54 male and 41 female fetuses with Down's syndrome studied. A shortened femur was identified in 28/54 (52%) males compared with 19/41 (46%) affected females (NS). A thickened nuchal fold was identified in 19/54 (35%) of males vs. 20/41 (49%) of females. Renal pyelectasis was seen in 7/54 (13%) males and 8/41 (19%) females. A heart defect was seen in 8/54 (15%) males and 7/41 (17%) females. Ventriculomegaly was identified in 6/54 (11%) males and 3/41 (7%) females with Down's syndrome. There were no statistically significant differences in the incidence of the sonographic findings when male and female Down's fetuses were compared. Our data show that the criteria for evaluation of sonographic markers for the ***identification*** of second-trimester fetuses with Down's syndrome should be the same in male and female fetuses.

CONTROLLED TERM: Check Tags: Female; Male
Abnormalities, Multiple: PP, physiopathology
*Abnormalities, Multiple: US, ultrasonography
Adult
*Down Syndrome: US, ultrasonography
Femur: AB, abnormalities
Femur: US, ultrasonography
*Fetal Diseases: US, ultrasonography
Fetal Heart: AB, abnormalities
Fetal Heart: US, ultrasonography
Heart Ventricles: AB, abnormalities
Heart Ventricles: US, ultrasonography
Humans
Humerus: AB, abnormalities
Humerus: US, ultrasonography
Karyotyping
Kidney: AB, abnormalities
Kidney: US, ultrasonography
Neck: AB, abnormalities
Neck: US, ultrasonography

Pregnancy
Pregnancy Trimester, Second
*Sex Characteristics
*Ultrasonography, Prenatal

L70 ANSWER 22 OF 25 HCAPLUS COPYRIGHT 2008 ACS on STN
ACCESSION NUMBER: 1994:506046 HCAPLUS Full-text
DOCUMENT NUMBER: 121:106046
ORIGINAL REFERENCE NO.: 121:19101h,19103a,19105a
ENTRY DATE: Entered STN: 03 Sep 1994
TITLE: Antenatal screening for chromosomal abnormalities
INVENTOR(S): Davies, Christopher John
PATENT ASSIGNEE(S): Kodak Ltd., UK; Eastman Kodak Co.
SOURCE: PCT Int. Appl., 26 pp.
CODEN: PIXXD2
DOCUMENT TYPE: Patent
LANGUAGE: English
INT. PATENT CLASSIF.:
MAIN: G01N033-76
SECONDARY: G01N033-68; G01N033-74
CLASSIFICATION: 14-13 (Mammalian Pathological Biochemistry)
Section cross-reference(s): 9
FAMILY ACC. NUM. COUNT: 1
PATENT INFORMATION:

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
WO 9412884	A1	19940609	WO 1993-EP3296	19931124
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W: JP, US				
RW: AT, BE, CH, DE, DK, ES, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE				
EP 627082	A1	19941207	EP 1994-901864	19931124
			<--	
EP 627082	B1	20000322		
EP 627082	B2	20061108		
R: AT, BE, CH, DE, DK, FR, GB, IT, LI, NL, SE				
JP 07503549	T	19950413	JP 1994-512756	19931124
			<--	
AT 191089	T	20000415	AT 1994-901864	19931124
			<--	
US 6010912	A	20000104	US 1995-566467	19951204
			<--	
JP 2005017305	A	20050120	JP 2004-239358	20040819
			<--	

PRIORITY APPLN. INFO.: GB 1992-24965 A

199211
28
<--
JP 1994-512756 A3
199311
24
<--
WO 1993-EP3296 W
199311
24
<--

PATENT CLASSIFICATION CODES:

PATENT NO.	CLASS	PATENT FAMILY CLASSIFICATION CODES
WO 9412884	ICM	G01N033-76
	ICS	G01N033-68; G01N033-74
	IPCI	G01N0033-76 [ICM,5]; G01N0033-68 [ICS,5]; G01N0033-74 [ICS,5]
	IPCR	G01N0033-50 [I,C*]; G01N0033-50 [I,A]; G01N0033-53 [I,C*]; G01N0033-53 [I,A]; G01N0033-68 [I,C*]; G01N0033-68 [I,A]; G01N0033-74 [I,C*]; G01N0033-74 [I,A]; G01N0033-76 [I,A]
	ECLA	G01N033/68T; G01N033/74; G01N033/74B; G01N033/76; S01N; S01N
EP 627082	IPCI	G01N0033-74 [I,C]; G01N0033-68 [I,C]; G01N0033-76 [I,A]; G01N0033-68 [I,A]; G01N0033-74 [I,A]
	IPCR	G01N0033-50 [I,C*]; G01N0033-53 [I,C*]; G01N0033-50 [I,A]; G01N0033-53 [I,A]
	ECLA	G01N033/68T; G01N033/74; G01N033/74B; G01N033/76; S01N; S01N
JP 07503549	IPCI	G01N0033-68 [ICM]; G01N0033-50 [ICS]
AT 191089	IPCI	G01N0033-76 [ICM,7]; G01N0033-68 [ICS,7]; G01N0033-74 [ICS,7]
	IPCR	G01N0033-50 [I,C*]; G01N0033-50 [I,A]; G01N0033-53 [I,C*]; G01N0033-53 [I,A]; G01N0033-68 [I,C*]; G01N0033-68 [I,A]; G01N0033-74 [I,C*]; G01N0033-74 [I,A]; G01N0033-76 [I,A]
	ECLA	G01N033/68T; G01N033/74; G01N033/74B; G01N033/76
US 6010912	IPCI	G01N0033-68 [ICM,6]
	IPCR	G01N0033-50 [I,C*]; G01N0033-50 [I,A]; G01N0033-53 [I,C*]; G01N0033-53 [I,A]; G01N0033-68 [I,C*]; G01N0033-68 [I,A]; G01N0033-74 [I,C*]; G01N0033-74 [I,A]; G01N0033-76 [I,A]
	NCL	436/510.000; 436/065.000; 436/086.000; 436/087.000; 436/811.000; 436/817.000; 436/818.000; 705/002.000
	ECLA	G01N033/68T; G01N033/74; G01N033/74B; G01N033/76; S01N; S01N
JP 2005017305	IPCI	G01N0033-53 [ICM]; G01N0033-76 [ICS]; G01N0033-74 [ICS,C*]
	IPCR	G01N0033-68 [I,A]; G01N0033-68 [I,C*]; G01N0033-74 [I,A]; G01N0033-74 [I,C*]; G01N0033-76 [I,A]
	ECLA	G01N033/68T; G01N033/74; G01N033/74B; G01N033/76
	FTERM	2G045/AA25; 2G045/AA27; 2G045/CA25; 2G045/CA26; 2G045/DA36; 2G045/DA54; 2G045/DA55; 2G045/JA01

ABSTRACT:

A method for antenatal screening for chromosomal ***abnormalities*** (in which maternal blood from a pregnant woman is measured for levels of free β hCG and at least a second serum ***marker*** and/or precursors and metabolites of these markers and the measured levels of these markers together with the gestational age of the pregnant woman are compared to reference values at various gestational ages of the levels for free β hCG and the second serum marker in (a) pregnant women carrying ***fetuses*** having abnormalities subject to the screen and (b) pregnant women carrying normal fetuses, the comparison being indicative of the risk of the pregnant woman carrying a ***fetus*** with an abnormality subject to the screen) is characterized in that the second serum marker is pregnancy-associated plasma protein A (PAPPA) and the screen is carried out by the end of the 13th completed week of pregnancy. An assay kit and an apparatus for the screening are also disclosed. When free β hCG and PAPPA were combined as serum markers there was significant improvement in detection rates for Down 's Syndrome.

SUPPL. TERM: pregnancy screening
chromosome abnormality blood
marker; chorionic gonadotropin beta
chromosome abnormality fetus
; PAPPA protein screening chromosome
abnormality fetus

INDEX TERM: Down's syndrome
Turner syndrome
(antenatal screening for, free β hCG and pregnancy-associated plasma protein A detn . in maternal human blood by end of week thirteen of pregnancy in)

INDEX TERM: Pregnancy
(free β hCG and pregnancy-associated plasma protein A determination in maternal human blood by end of week thirteen of, in antenatal screening for chromosomal abnormalities in fetus)

INDEX TERM: Blood analysis
(free β hCG and pregnancy-associated plasma protein A determination in maternal human, in antenatal screening for chromosomal abnormalities)

INDEX TERM: Computers
(in apparatus for determination of free β hCG and pregnancy-associated plasma protein A in maternal human blood by end of week thirteen of pregnancy, antenatal screening for chromosomal abnormalities in fetus in relation to)

INDEX TERM: Trisomy syndrome
(18, antenatal screening for, free β hCG and pregnancy-associated plasma protein A determination in maternal human blood by end of week thirteen of pregnancy in)

INDEX TERM: Testis, disease
(Klinefelter's syndrome, antenatal screening for, free β hCG and pregnancy-associated plasma protein A determination

in maternal human blood by end of week thirteen of pregnancy in)

INDEX TERM: Sialoglycoproteins
(PAPP-A (pregnancy-associated plasma protein A), determination in maternal human blood of, in antenatal screening for chromosomal abnormalities in fetus)

INDEX TERM: Trisomy syndrome
(Patau's syndrome, antenatal screening for, free β hCG and pregnancy-associated plasma protein A determination in maternal human blood by end of week thirteen of pregnancy in)

INDEX TERM: Analysis
(apparatus, for free β hCG and pregnancy-associated plasma protein A determination in maternal human blood by end of week thirteen of pregnancy, antenatal screening for chromosomal abnormalities in fetus in relation to)

INDEX TERM: Chromosome
(disease, abnormalities, antenatal screening for, free β hCG and pregnancy-associated plasma protein A determination in maternal human blood by end of week thirteen of pregnancy in)

INDEX TERM: Embryo
(fetus, chromosomal abnormalities in, antenatal screening for, free β hCG and pregnancy-associated plasma protein A determination in maternal human blood by end of week thirteen of pregnancy in)

INDEX TERM: Fetoproteins
(α-, determination in maternal human blood of, in antenatal screening for chromosomal abnormalities in fetus)

INDEX TERM: 57-83-0, Progesterone, analysis 651-48-9, Dehydroepiandrosterone sulfate 4873-65-8, 16α-Hydroxydehydroepiandrosterone 3-sulfate
ROLE: ANT (Analyte); ANST (Analytical study)
(determination in maternal human blood of, in antenatal screening for chromosomal abnormalities in fetus)

INDEX TERM: 57285-09-3, Inhibin
ROLE: BAC (Biological activity or effector, except adverse); BSU (Biological study, unclassified); BIOL (Biological study)
(determination in maternal human blood of, in antenatal screening for chromosomal abnormalities in fetus)

INDEX TERM: 9002-61-3, Chorionic gonadotropin
ROLE: BAC (Biological activity or effector, except adverse); BSU (Biological study, unclassified); BIOL (Biological study)
(free β chain of, determination in maternal

human blood of, in antenatal screening for chromosomal abnormalities in fetus)

INDEX TERM: 50-27-1, Estriol
ROLE: BAC (Biological activity or effector, except adverse); BSU (Biological study, unclassified); BIOL (Biological study)
(unconjugated, determination in maternal human blood of, in antenatal screening for chromosomal abnormalities in fetus)

L70 ANSWER 23 OF 25 BIOSIS COPYRIGHT (c) 2008 The Thomson Corporation on STN DUPLICATE 4
ACCESSION NUMBER: 1993:385849 BIOSIS Full-text
DOCUMENT NUMBER: PREV199396061149
TITLE: Biparietal diameter and crown-rump length in fetuses with Down's syndrome: Implications for antenatal serum screening for Down's syndrome.
AUTHOR(S): Wald, N. J. [Reprint author]; Smith, D.; Kennard, A.; Palomaki, G. E.; Salonen, R.; Holzgreve, W.; Pejtsik, B.; Coombes, E. J.; Mancini, G.
CORPORATE SOURCE: Dep. Environmental Preventive Med., Wolfson Inst. Preventive Med., Med. Coll. St. Bartholomew's Hosp., London EC1M 6BQ, UK
SOURCE: British Journal of Obstetrics and Gynaecology, (1993) Vol. 100, No. 5, pp. 430-435.
CODEN: BJOGAS. ISSN: 0306-5456.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 23 Aug 1993
Last Updated on STN: 23 Aug 1993
ABSTRACT:Objectives: 1. To compare the ultrasound biparietal diameter and crown-rump length of fetuses with and without Down's syndrome in the first half of pregnancy; 2. To investigate the effect of estimation of gestational age using either measure on the detection rate of serum screening for Down's ***syndrome.*** Design: Matched case-control study. Cases were singleton Down's syndrome pregnancies with a biparietal diameter or a crown-rump length recorded. Five controls were matched to each case on: medical centre; the data of the ultrasound scan examination (within two years); gestational age measured as the number of days since the first day of the last menstrual period; and the ultrasound measure used (ie the biparietal diameter (the measure of choice), or the crown-rump length otherwise). If a woman had a serum ***screening*** test for Down's syndrome, the biparietal diameter or crown-rump length measurement had to be taken prior to the screening test so that the result of the test could not influence whether a scan was performed. Setting: Ten antenatal screening centres in seven countries in Europe and North America. Subjects: Two hundred and one women with singleton Down's syndrome ***pregnancies*** and 1005 women with unaffected singleton pregnancies. Results: The median biparietal diameter of fetuses with ***Down*** 's syndrome was identical to that among the controls (median difference 0.0 mm, 95% confidence intervals (CI) -0.5 to 0.5 mm). The estimates of gestational age based on biparietal diameter yielded a median gestational age less than that based on the women's last menstrual period: three days less for cases and two days less for

controls; small but statistically significant differences probably reflected a minor systematic difference in the conversion of a biparietal diameter to a gestational age estimate. The median crown-rump length of fetuses with Down's syndrome was also identical to that among controls (median difference 0.0 mm, 95% CI -1.5 to 2.0 mm). There was no significant difference between the median gestational age estimate based on crown-rump length and that based on the women's last menstrual period. Conclusion: In antenatal ***screening*** for Down's syndrome the routine use of an ultrasound biparietal diameter or crown-rump length measurement to estimate gestational age will not adversely affect the detection rate. To avoid differences in gestational age estimates using the last menstrual period and the biparietal diameter influencing screening performance, separate medians should be derived for each serum ***marker*** using the two methods of estimating gestational age. The appropriate set of medians can then be used to calculate the multiple of the median value for each woman screened depending on the method used to estimate her gestational age.

CONCEPT CODE: Genetics - Human 03508
Radiation biology - Radiation and isotope techniques 06504
Physiology - General 12002
Pathology - Diagnostic 12504
Blood - Blood and lymph studies 15002
Reproductive system - Physiology and biochemistry 16504
Nervous system - Pathology 20506
Psychiatry - Mental retardation 21006
Development and Embryology - Descriptive teratology and teratogenesis 25552

INDEX TERMS: Major Concepts
Development; Neurology (Human Medicine, Medical Sciences); Pathology; Physiology; Psychiatry (Human Medicine, Medical Sciences); Reproductive System (Reproduction)

INDEX TERMS: Miscellaneous Descriptors
HYPOXIA; RESPIRATORY DISTRESS SYNDROME

ORGANISM: Classifier
Hominidae 86215
Super Taxa
Primates; Mammalia; Vertebrata; Chordata; Animalia
Organism Name
human
Taxa Notes
Animals, Chordates, Humans, Mammals, Primates, Vertebrates

L70 ANSWER 24 OF 25 MEDLINE on STN
ACCESSION NUMBER: 1993148344 MEDLINE Full-text
DOCUMENT NUMBER: PubMed ID: 1283415
TITLE: Sonographic scoring index for prenatal detection of chromosomal abnormalities.
AUTHOR: Benacerraf B R; Neuberg D; Bromley B; Frigoletto F D Jr
CORPORATE SOURCE: Department of Obstetrics & Gynecology, Brigham & Women's Hospital, Boston, Massachusetts.
SOURCE: Journal of ultrasound in medicine : official journal of the American Institute of Ultrasound in Medicine, (1992 Sep) Vol. 11, No. 9, pp. 449-58.

Journal code: 8211547. ISSN: 0278-4297.
PUB. COUNTRY: United States
DOCUMENT TYPE: (COMPARATIVE STUDY)
Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 199303
ENTRY DATE: Entered STN: 12 Mar 1993
Last Updated on STN: 25 Jan 2002
Entered Medline: 1 Mar 1993

ABSTRACT:
Current indications for cytogenetic evaluation leave the majority of ***Down*** ***syndrome*** ***fetuses*** undetected. Using advanced maternal age and low maternal serum alpha-fetoprotein (AFP) levels as criteria, only 40% of ***fetuses*** with ***Down*** ***syndrome*** (trisomy 21) are ***identified*** (positive predictive ***value***, 0.4% to 1%). We evaluate the sonographically detectable physical features of ***second*** trimester ***fetuses*** to determine whether these features are more sensitive and specific than maternal age for detecting ***fetuses*** with abnormal karyotypes. From March 1, 1990, to September 1, 1991, more than 5,000 ***fetuses*** between 14 and 20 weeks of development were referred for genetic amniocentesis because of advanced maternal age or abnormal AFP levels. Forty-three of these 5,000 ***fetuses*** were later found to have autosomal trisomies by karyotype (32 with trisomy 21, nine with trisomy 18, and two with trisomy 13). A sample of 588 consecutive normal ***fetuses*** from the total of more than 5,000 amniocenteses performed during this period of time was used as our control group for ***statistical*** analysis. The sonographic features of these 588 normal ***second*** trimester ***fetuses*** and the 43 trisomic ***fetuses*** recorded prospectively prior to knowledge of the karyotype were evaluated ***statistically***. The femur and humerus lengths, nuchal fold, renal pelvic dimension, and major structural defects were compared in the normal and trisomic ***fetuses.*** On the basis of our results, a weighted sonographic score was developed to optimize the detection of ***fetuses*** at risk for aneuploidy. Using our previously published formulas and criteria for a short femur and humerus, 17/32 (53%) ***fetuses*** with ***Down*** ***syndrome*** and 23/588 (3.9%) of the normal ***fetuses*** were identified. Twenty two of 32 ***Down*** ***syndrome*** ***fetuses*** (69%) and 2/588 (0.34%) of normals had a nuchal fold > or = 6 mm, and 11 of 32 ***Down*** ***syndrome*** ***fetuses*** and all those with trisomies 18 and 13 had a major anomaly detected sonographically. The following scoring system was developed for the detection of aneuploidy: nuchal fold = 2, major structural defect = 2, and short femur, short humerus, and pyelectasis = 1 each. Selecting ***fetuses*** with a score of > or = 2 would ***identify*** 26/32 (81%) ***Down*** ***syndrome*** ***fetuses***, and 9/9 (100%) and 2/2 (100%) ***fetuses*** with trisomies 18 and 13 respectively, but only 26/588 (4.4%) of the normal ***fetuses.*** Using the sonographic score of 2 results in a positive predictive ***value*** for a 1/250 risk group of 6.87% for ***identifying*** ***Down*** ***syndrome*** ***fetuses*** and 7.25% for all three trisomies.(ABSTRACT TRUNCATED AT 400 WORDS)

CONTROLLED TERM: Check Tags: Female
Amniocentesis
Aneuploidy
*Chromosome Aberrations: US, ultrasonography
Chromosome Disorders
Chromosomes, Human, Pair 13

Chromosomes, Human, Pair 18
Down Syndrome: US, ultrasonography
*Fetal Diseases: US, ultrasonography
Gestational Age
Humans
Karyotyping
Predictive Value of Tests
Pregnancy
Prospective Studies
Sensitivity and Specificity
Trisomy
*Ultrasonography, Prenatal: MT, methods
alpha-Fetoproteins: AN, analysis
CHEMICAL NAME: 0 (alpha-Fetoproteins)

L70 ANSWER 25 OF 25 MEDLINE on STN
ACCESSION NUMBER: 1990084629 MEDLINE Full-text
DOCUMENT NUMBER: PubMed ID: 2480649
TITLE: Down's syndrome: current screening techniques.
AUTHOR: White R S 3rd
CORPORATE SOURCE: Department of Obstetrics and Gynecology, George Washington University Medical Center, Washington, DC.
SOURCE: Southern medical journal, (1989 Dec) Vol. 82, No. 12, pp. 1483-6.
Journal code: 0404522. ISSN: 0038-4348.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Abridged Index Medicus Journals; Priority Journals
ENTRY MONTH: 199001
ENTRY DATE: Entered STN: 28 Mar 1990
Last Updated on STN: 3 Feb 1997
Entered Medline: 23 Jan 1990
ABSTRACT:
Antenatal screening for Down's syndrome traditionally relied upon performing amniocentesis for karyotype on pregnant women aged 35 years and older. This method detects approximately 20% of all Down's syndrome ***pregnancies*** , with a false-positive rate of 4.3%. By incorporating maternal serum alpha-fetoprotein values as an additional screening parameter to maternal age, 28% of all Down's syndrome pregnancies may be ***diagnosed*** , with a 35% reduction in false-positive results. Other ***screening*** parameters such as maternal serum unconjugated estriol and human chorionic gonadotropin may eventually make it possible to detect more than 65% of pregnancies with ***chromosomally*** abnormal fetuses, without compromise in false-positive rates.
CONTROLLED TERM: Check Tags: Female
Amniocentesis
Down Syndrome: BL, blood
*Down Syndrome: DI, diagnosis
False Positive Reactions
Fetal Diseases: BL, blood
*Fetal Diseases: DI, diagnosis
Humans
*Maternal Age
Pregnancy
*Prenatal Diagnosis: MT, methods

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                    Prenatal Diagnosis: ST, standards
                      Probability
                    *Reagent Kits, Diagnostic: ST, standards
                    Risk Factors
                    *alpha-Fetoproteins: AN, analysis
CHEMICAL NAME:      0 (Reagent Kits, Diagnostic); 0 (alpha-Fetoproteins)


=> d his nofile

     (FILE 'HOME' ENTERED AT 14:39:30 ON 15 DEC 2008)

     FILE 'HCAPLUS' ENTERED AT 14:39:42 ON 15 DEC 2008
L1             1 SEA ABB=ON  PLU=ON  US20070148631/PN
               SEL RN

     FILE 'REGISTRY' ENTERED AT 14:40:15 ON 15 DEC 2008
L2             4 SEA ABB=ON  PLU=ON  (102510-92-9/BI OR 151662-33-8/BI OR
               50-27-1/BI OR 9002-61-3/BI)
               D SCA

     FILE 'WPIX' ENTERED AT 14:40:25 ON 15 DEC 2008
L3             1 SEA ABB=ON  PLU=ON  US20070148631/PN
               D SCA
               D IFULL

     FILE 'HCAPLUS' ENTERED AT 15:09:42 ON 15 DEC 2008
               E CHROMOSOME ABERRATIONS/CT
L4         15891 SEA ABB=ON  PLU=ON  "CHROMOSOME ABERRATIONS"+PFT,NT/CT
               E DOWNS SYNDROME/CT
               E "DOWN'S SYNDROME"/CT
L5          3715 SEA ABB=ON  PLU=ON  "DOWN'S SYNDROME"+PFT,NT/CT
               E PREGNANCY/CT
L6         70345 SEA ABB=ON  PLU=ON  PREGNANCY+PFT,NT/CT
L7           508 SEA ABB=ON  PLU=ON  L6 AND (L4 OR L5)
L8               QUE ABB=ON  PLU=ON  DETERMIN? OR IDENTIF? OR DIAGNOS? OR
               DETECT?
L9               QUE ABB=ON  PLU=ON  SCREEN?
L10          431 SEA ABB=ON  PLU=ON  L7 AND (L8 OR L9)
L11              QUE ABB=ON  PLU=ON  FETUS
L12          219 SEA ABB=ON  PLU=ON  L10 AND L11
L13              QUE ABB=ON  PLU=ON  CHROMOSOM?(2A)ABNORMAL?
L14              QUE ABB=ON  PLU=ON  DOWN(2A)SYNDROME?
L15         1450 SEA ABB=ON  PLU=ON  (L8 OR L9)(3A)(L13 OR L14)
L16          108 SEA ABB=ON  PLU=ON  L12 AND L15
L17              QUE ABB=ON  PLU=ON  MARKER? OR INDICAT!R?
L18              QUE ABB=ON  PLU=ON  PARAMETER? OR VALUE
L19           71 SEA ABB=ON  PLU=ON  L16 AND (L17 OR L18)
L20       314761 SEA ABB=ON  PLU=ON  (L8 OR L9)(5A)(L17 OR L18)
L21           42 SEA ABB=ON  PLU=ON  L19 AND L20
L22              QUE ABB=ON  PLU=ON  (PREGNAN? OR FETUS)(3A)(L13 OR L14)
L23           29 SEA ABB=ON  PLU=ON  L21 AND L22
L24              QUE ABB=ON  PLU=ON  FIRST? OR 1ST OR 1(W)ST
L25              QUE ABB=ON  PLU=ON  SECOND? OR 2ND OR 2(W)ND
L26           11 SEA ABB=ON  PLU=ON  L23 AND L24
L27           15 SEA ABB=ON  PLU=ON  L23 AND L25
L28            5 SEA ABB=ON  PLU=ON  L26 AND L27
               D KWIC 1-2
L29           26 SEA ABB=ON  PLU=ON  L23 AND (PY<=2006 OR PRY<=2006 OR
               AY<=2006)
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L30             QUE ABB=ON  PLU=ON  STATIST? OR COMPUTER? OR PROGRAM?
L31           8 SEA ABB=ON  PLU=ON  L29 AND L30

     FILE 'WPIX' ENTERED AT 16:03:01 ON 15 DEC 2008
L32         307 SEA ABB=ON  PLU=ON  (L8 OR L9)(3A)(L13 OR L14)
L33          49 SEA ABB=ON  PLU=ON  L32 AND L11
L34          23 SEA ABB=ON  PLU=ON  L33 AND (L17 OR L18)
L35          18 SEA ABB=ON  PLU=ON  L34 AND L22
L36          18 SEA ABB=ON  PLU=ON  L35 AND (PY<=2006 OR PRY<=2006 OR
                AY<=2006)

     FILE 'BIOSIS' ENTERED AT 16:14:20 ON 15 DEC 2008
L37        3277 SEA ABB=ON  PLU=ON  (L8 OR L9)(3A)(L13 OR L14)
L38         596 SEA ABB=ON  PLU=ON  L37 AND L11
L39         252 SEA ABB=ON  PLU=ON  L38 AND (L17 OR L18)
L40         128 SEA ABB=ON  PLU=ON  L39 AND L22
L41          56 SEA ABB=ON  PLU=ON  L40 AND L20
                D KWIC 1-2
L42             QUE ABB=ON  PLU=ON  PROBABILIT?
L43             QUE ABB=ON  PLU=ON  STATISTIC?
L44           6 SEA ABB=ON  PLU=ON  L41 AND (L42 OR L43)
L45           5 SEA ABB=ON  PLU=ON  L44 AND PY<=2006

     FILE 'EMBASE' ENTERED AT 16:18:11 ON 15 DEC 2008
L46        3752 SEA ABB=ON  PLU=ON  (L8 OR L9)(3A)(L13 OR L14)
L47        1402 SEA ABB=ON  PLU=ON  L46 AND L11
L48         716 SEA ABB=ON  PLU=ON  L47 AND (L17 OR L18)
L49         294 SEA ABB=ON  PLU=ON  L48 AND L22
L50         167 SEA ABB=ON  PLU=ON  L49 AND L20
L51          25 SEA ABB=ON  PLU=ON  L50 AND (L42 OR L43)
L52          22 SEA ABB=ON  PLU=ON  L51 AND PY<=2006
                D SCA
L53           9 SEA ABB=ON  PLU=ON  L52 AND L24
L54          15 SEA ABB=ON  PLU=ON  L52 AND L25
L55           6 SEA ABB=ON  PLU=ON  L53 AND L54

     FILE 'MEDLINE' ENTERED AT 16:26:50 ON 15 DEC 2008
L56        3989 SEA ABB=ON  PLU=ON  (L8 OR L9)(3A)(L13 OR L14)
L57         704 SEA ABB=ON  PLU=ON  L56 AND L11
L58         385 SEA ABB=ON  PLU=ON  L57 AND (L17 OR L18)
L59         225 SEA ABB=ON  PLU=ON  L58 AND L22
L60          87 SEA ABB=ON  PLU=ON  L59 AND L20
L61          15 SEA ABB=ON  PLU=ON  L60 AND (L42 OR L43)
L62          12 SEA ABB=ON  PLU=ON  L61 AND PY<=2006
L63           4 SEA ABB=ON  PLU=ON  L62 AND L24
L64           7 SEA ABB=ON  PLU=ON  L62 AND L25
L65           2 SEA ABB=ON  PLU=ON  L63 AND L64
L66          12 SEA ABB=ON  PLU=ON  (L62 OR L63 OR L64 OR L65)

     FILE 'WPIX' ENTERED AT 16:29:07 ON 15 DEC 2008
L67           4 SEA ABB=ON  PLU=ON  L36 AND (L42 OR L43)
                SEL L67 PN,AP

     FILE 'HCAPLUS' ENTERED AT 16:30:37 ON 15 DEC 2008
L68           5 SEA ABB=ON  PLU=ON  (WO1993-US7408/AP OR EP1990-903086/AP

L69           7 SEA ABB=ON  PLU=ON  L31 NOT L68

     FILE 'HCAPLUS, BIOSIS, EMBASE, MEDLINE' ENTERED AT 16:31:13 ON 15
     DEC 2008
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L70          25 DUP REM L69 L45 L55 L66 (5 DUPLICATES REMOVED)


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